



UNIVERSIDADE DE LISBOA
FACULDADE DE MOTRICIDADE HUMANA



Gait Analyses of a Portuguese Child with *Cerebroretinal Microangiopathy with Calcifications and Cysts* under specific walking conditions: barefoot, orthoses with shoe, insole with shoe

A Case Study

Dissertação com vista à obtenção do Grau de Mestre na Especialidade de
Ciências da Fisioterapia

Orientador:

Doutor António Prieto Veloso

Júri:

Doutor António Prieto Veloso (Presidente do Júri)

Doutora Maria Salomé Silva de Almeida (Vogal)

Doutora Maria Filomena Araújo Costa Cruz Carnide (Vogal)

Catarina Rosa Francisco Rodrigues Góis

2014



UNIVERSIDADE DE LISBOA
FACULDADE DE MOTRICIDADE HUMANA



Gait Analyses of a Portuguese Child with *Cerebroretinal Microangiopathy with Calcifications and Cysts* under specific walking conditions: barefoot, orthoses with shoe, insole with shoe

A Case Study

Dissertação com vista à obtenção do Grau de Mestre na Especialidade de
Ciências da Fisioterapia

Orientador:

Doutor António Prieto Veloso

Júri:

Doutor António Prieto Veloso (Presidente do Júri)

Doutora Maria Salomé Silva de Almeida (Vogal)

Doutora Maria Filomena Araújo Costa Cruz Carnide (Vogal)

Catarina Rosa Francisco Rodrigues Góis

2014

ACKNOWLEDGEMENTS

First of all I would like to acknowledge Prof. Dr. António Veloso for the opportunity to develop my master thesis in the Biomechanics and Functional Morphology Laboratory, with a wonderful team that is coordinated by him, accepting me as his student, supervising and giving me all the conditions needed to develop my master.

A special thanks to Prof. Raul Oliveira who has supported me since I began this journey, showing me the positive side of this journey and being so present.

Then I would like to extend this acknowledgement to all my master colleagues who teach me a lot and support me in many ways, and to the BFML team, always available to support and teach me.

To all the master teachers and professionals involved (in)directly in my master, also the parents and of course the child... all of you have been very important and I am extremely grateful for having your support and your contribution in my growth process.

My family has been so important that I cannot find words to say how grateful I am, how much I feel your support and how important that is to me. To all of you (mum, dad, brother, boyfriend, sister-in-law) THANK YOU SO MUCH.

Finally, to my niece Maria, the youngest family member, but the one that gives me more motivation and courage, you are such an inspiration.

ABSTRACT

The objective of this case study was to describe and compare differences in gait patterns of a child suffering from *Cerebroretinal Microangiopathy with Calcifications and Cysts*, along the time (three data collections) under different conditions (barefoot, foot orthoses and insole).

We performed a biomechanical analysis collecting spatio-temporal parameters and kinematic data, at free walking speed, during his 4th and 5th years of life.

Different conditions were compared to verify the efficiency of the foot orthoses and insoles prescribed by doctors who assist him, and the effects over barefoot walking.

Using an adapted set-up of markers based on Oxford multi-segmented foot, adapted to the child's condition, data collection took place at the Biomechanics and Functional Morphology Laboratory, with 14 infrared cameras Qualisys Oqus 300, the collection was made with Qualisys Track Manager, and data was processed with Visual 3D. Data were collected according to the adaptation or prescription of foot orthoses, after a period of adaptation to their use.

In each data collection, it was observed that new adaptations in foot orthoses benefit ankle movement under the three planes and adaptations in the lower limb joints. However, the effect of other kind of stimulus, such as physiotherapy, is unknown, as well as the direct effects of the disease evolution under his gait pattern, due to neural plasticity, loss of vision, individual's ability to adapt to conditions.

By maintaining regular data collection, it will be possible to contribute with more information about this rare and poorly described disease

development, clarifying the importance of different factors influence under his gait patterns, as well as contributing to adaptations that will benefit him and improve his quality of life.

Key words: Gait, Kinematics, Spatio-temporal parameters, *Cerebroretinal Microangiopathy with Calcifications and Cysts*, Foot Orthoses, Insoles, Barefoot

TABLE OF CONTENTS

CHAPTER 1 – INTRODUCTION.....	1
1. Introduction.....	1
2. Literature Review.....	4
2.1. Introduction	4
2.2. <i>Cerebroretinal Microangiopathy with Calcifications and Cysts</i>	6
2.3. Neural development and plasticity	8
2.3. Visual Impairment.....	14
2.4. Gait cycle	19
2.4.1. Spatial Reference System.....	20
2.4.2. Gait Cycle Spatio-Temporal Parameters.....	20
2.4.3. Gait Cycle Kinematics	23
2.4.3.1. Gait Cycle Description over Sagittal Plane	24
2.4.3.1.1. Ankle	24
2.4.3.1.2. Knee	27
2.4.3.1.3. Hip	29
2.4.3.1.4. Pelvis.....	30
2.4.3.2. Gait Cycle Description over Frontal Plane	31
2.4.3.2.1. Ankle	31
2.4.3.2.2. Knee	31
2.4.3.2.3. Hip	32
2.4.3.2.4. Pelvis.....	33
2.4.3.3. Gait Cycle Description over Transverse Plane	34
2.4.3.3.1. Ankle	35
2.4.3.3.2. Knee	35
2.4.3.3.3. Hip	36
2.4.3.3.4. Pelvis.....	37
2.5. Considerations about children gait cycle	39
2.6. Foot Orthoses	42
2.6.1. Ankle Foot Orthoses	44
2.6.2. Insole	45
CHAPTER 2 – METHODS AND MATERIALS.....	46
1. Methodology	46

1.1. Study Type	46
1.2. Subject History	47
3.3. Instruments for data collection and procedures.....	54
3.3.1. Information collected from medical reports and by interview	54
3.3.2. Instruments for Kinematic and Anthropometric data collection	55
3.3.3. Data Collection.....	58
3.3.3. Anatomical Points	60
3.3.4. Calibration.....	63
3.3.5. Data Processing.....	65
3.4. Variables Operational Definition.....	68
CHAPTER 3 – RESULTS & DISCUSSION	69
1. Results Presentation and Discussion	69
1.1. 1 st Data Collection: 11.01.2013	72
1.1.1. Temporal Distance Results ($X \pm \delta$)	72
1.1.2. Intersegmental angle on sagittal plane	74
1.1.2.1. Ankle.....	74
1.1.2.2. Knee	76
1.1.2.3. Hip	78
1.1.2.4. Pelvis	79
1.1.3. Intersegmental angles on frontal plane	81
1.1.3.1. Ankle.....	81
1.1.3.2. Knee	82
1.1.3.3. Hip	84
1.1.3.4. Pelvis	85
1.1.4. Intersegmental angles in transverse plane	86
1.1.4.1. Ankle.....	86
1.1.4.2. Knee	87
1.1.4.3. Hip	88
1.1.4.4. Pelvis	89
1.2. 2 nd Data Collection: 22.03.2013	90
1.2.1. Temporal Distance Results ($X \pm \delta$)	90
1.2.2. Intersegmental angle on sagittal plane	92
1.2.2.1. Ankle.....	92

1.2.2.2. Knee	94
1.2.2.3. Hip	96
1.2.2.4. Pelvis	97
1.2.3. Intersegmental angles on frontal plane	99
1.2.3.1. Ankle.....	99
1.2.3.2. Knee	100
1.2.3.3. Hip	102
1.2.3.4. Pelvis	103
1.2.4. Intersegmental angles in transverse plane	104
1.2.4.1. Ankle.....	104
1.2.4.2. Knee	105
1.2.4.3. Hip	106
1.3. 3 rd Data Collection: 19.07.2013	108
1.3.1. Temporal Distance Results ($X \pm \delta$):	108
1.3.2. Intersegmental angle on sagittal plane	110
1.3.2.1. Ankle.....	110
1.3.2.2. Knee	112
1.3.2.3. Hip	114
1.3.2.4. Pelvis	116
1.3.3. Intersegmental angles on frontal plane	117
1.3.3.1. Ankle.....	117
1.3.3.2. Knee	118
1.3.3.3. Hip	120
1.3.3.4. Pelvis	121
1.3.4. Intersegmental angles in transverse plane	122
1.3.4.1. Ankle.....	122
1.3.4.2. Knee	123
1.3.4.3. Hip	124
1.3.4.4. Pelvis	125
1.4. Barefoot and Orthoses with shoe conditions along time.....	127
1.4.1. Barefoot Condition	127
1.4.1.1. Temporal Distance Results ($X \pm \delta$)	127
1.4.1.2. Intersegmental Angles on Sagittal Plane	129
1.4.1.2.1. Ankle	129

1.4.1.2.2. Knee	130
1.4.1.2.3. Hip	131
1.4.1.2.4. Pelvis	131
1.4.1.3. Intersegmental Angles on Frontal Plane	132
1.4.1.3.1. Ankle	132
1.4.1.3.2. Knee	133
1.4.1.3.3. Hip	134
1.4.1.3.4. Pelvis	134
1.4.1.3. Intersegmental Angles on Transverse Plane	135
1.4.1.3.1. Ankle	135
1.4.1.3.2. Knee	135
1.4.1.3.3. Hip	136
1.4.1.3.4. Pelvis	137
1.4.2. Orthoses with shoe condition	138
1.4.2.1. Temporal Distance Results ($X \pm \delta$)	138
1.4.2.2. Intersegmental Angles on Sagittal Plane	139
1.4.2.2.1. Ankle	139
1.4.2.2.2. Knee	140
1.4.2.2.3. Hip	141
1.4.2.2.4. Pelvis	141
1.4.2.3. Intersegmental Angles on Frontal Plane	141
1.4.2.3.1. Ankle	141
1.4.2.3.2. Knee	142
1.4.2.3.3. Hip	143
1.4.2.3.4. Pelvis	143
1.4.2.4. Intersegmental Angles on Transverse Plane	143
1.4.2.4.1. Ankle	143
1.4.2.4.2. Knee	144
1.4.2.4.3. Hip	145
1.4.2.4.4. Pelvis	146
CHAPTER 4 – CONCLUSIONS	147
BIBLIOGRAPHY	150
APPENDIX A	154
APPENDIX B	158

APPENDIX C.....	171
APPENDIX D.....	175

INDEX OF TABLES

Table 1 – Mean time/distance parameters for 5-year-old normal subjects	22
Table 2 – Temporal Distance Results 1 st Data Collection.....	72
Table 3 - Ankle intersegmental angle on sagittal plane (dorsiflexion [+], plantar flexion [-]) Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	74
Table 4 – Ankle velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.....	75
Table 5 - Knee intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	76
Table 6 - Knee velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.....	77
Table 7 - Hip intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	78
Table 8 - Hip velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.....	79
Table 9 - Pelvis intersegmental angle on sagittal plane (anterior flexion [+], posterior flexion [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.....	79
Table 10 - Ankle intersegmental angle on frontal plane (eversion [+], inversion [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	81

Table 11 - Knee intersegmental angle on frontal plane (adduction [+], abduction [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.	82
Table 12 - Hip intersegmental angle on frontal plane (abduction [+], adduction [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.	84
Table 13 – Pelvis intersegmental angle on frontal plane (Up [+], Down [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.	85
Table 14 - Ankle intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe.	86
Table 15 - Knee intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe.	87
Table 16 - Hip intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.	88
Table 17 - Pelvis intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.	89
Table 18 – Temporal Distance Results 2 nd Data Collection.....	90
Table 19 – Ankle intersegmental angle on sagittal plane (dorsiflexion [+], plantar flexion [-]). Green – Orthoses with shoe; Black – Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.	92
Table 20 – Ankle velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – Orthoses with shoe; Black – Barefoot.	93
Table 21 - Knee intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – Orthoses with shoe; Black - Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.	94
Table 22 - Knee velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – Orthoses with shoe; Black – Barefoot.....	95

Table 23 - Hip intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – Orthoses with shoe; Black - Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.	96
Table 24 - Hip velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – Orthoses with shoe; Black – Barefoot.....	97
Table 25 - Pelvis intersegmental angle on sagittal plane (anterior flexion [+], posterior flexion [-]). Green – Orthoses with shoe; Black – Barefoot.	97
Table 26 - Ankle intersegmental angle on frontal plane (eversion [+], inversion [-]). Green – Orthoses with shoe; Black – Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.	99
Table 27 - Knee intersegmental angle on frontal plane (adduction [+], abduction [-]). Green – Orthoses with shoe; Black – Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.	100
Table 28 - Hip intersegmental angle on frontal plane (abduction [+], adduction [-]). Green – Orthoses with shoe; Black - Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.	102
Table 29 – Pelvis intersegmental angle on frontal plane (Up [+], Down [-]). Green – Orthoses with shoe; Black – Barefoot.....	103
Table 30 - Ankle intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot.....	104
Table 31 - Knee intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot.....	105
Table 32 - Hip intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot.....	106
Table 33 - Pelvis intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot.....	107
Table 34 – Temporal Distance Results 3 rd Data Collection	108

Table 35 - Ankle intersegmental angle on sagittal plane (dorsiflexion [+], plantar flexion [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	110
Table 36 – Ankle velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.....	111
Table 37 - Knee intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	112
Table 38 - Knee velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.....	114
Table 39 - Hip intersegmental angle on sagittal plane (flexion [+], extension [-]).Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	114
Table 40 - Hip velocity on sagittal plane (velocity increase [+], velocity decrease [-]).Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.....	115
Table 41 - Pelvis intersegmental angle on sagittal plane (anterior flexion [+], posterior flexion [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.....	116
Table 42 - Ankle intersegmental angle on frontal plane (eversion [+], inversion [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	117
Table 43 - Knee intersegmental angle on frontal plane (adduction [+], abduction [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	118
Table 44- Hip intersegmental angle on frontal plane (abduction [+], adduction [-]).Green – orthoses with shoe; Black - Barefoot; Beige –	

insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.....	120
Table 45 – Pelvis intersegmental angle on frontal plane (Up [+], Down [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.....	121
Table 46 - Ankle intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.	122
Table 47 - Knee intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.	123
Table 48 - Hip intersegmental angle on transverse plane (internal rotation [+], external rotation [-]).Green – orthoses with shoe; Black – Barefoot; Beige – insole with shoe.	124
Table 49 - Pelvis intersegmental angle on transverse plane (internal rotation [+], external rotation [-]).Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.	125
Table 50 – Temporal Distance Results for Barefoot Condition.....	127
Table 51 – Temporal Distance Results for Orthoses with Shoe condition	138

INDEX OF PHOTOS

Figure 1 – Child photo static pose, posterior view (left) view between sagittal and frontal plane (right)	52
Figure 2 – Child photo static pose, lateral view (left) and feet (right) ...	52
Figure 3 – Laboratory View	56
Figure 4 – Reflective Spheres used.....	57
Figure 5 – Hall used for Data Collection	57
Figure 6 – Set up for barefoot condition	61
Figure 7 – Set up for orthoses with shoe condition.....	61
Figure 8 – Barefoot condition	69
Figure 9 – Orthoses image before getting into the shoe (left) and shoe (right)	69
Figure 10 – Shoe condition.....	70
Figure 11 – Insoles.....	70

CHAPTER 1 – INTRODUCTION

1. Introduction

The present study, which is a case study, is about the gait characterization of a 5-year-old boy, who suffers from *Cerebroretinal Microangiopathy with Calcifications and Cysts* (CRMCC).

There are few studies about *Cerebroretinal Microangiopathy with Calcifications and Cysts* because of low prevalence, and the few cases identified.

Cerebroretinal Microangiopathy with Calcifications and Cysts, is a rare syndrome described by some investigators based on study cases, or observational reports. This syndrome is characterized by cysts and calcifications that develop in the brain, associated with leukoencephalopathy and retinal vascular abnormalities (1).

There are few cases related in literature and published in journals with impact factor and even fewer studies published (none was found) about gait biomechanics analysis of the known cases.

The lack of articles about gait biomechanics analysis might be justified by evidence of variability between subjects with this diagnosis, but also for their unpredictable average life expectancy, and because most of the papers describe the subjects after their death according to the information collected during their lives.

The present study will focus on the biomechanics analysis of a child's gait with this diagnosis, describing gait spatio-temporal parameters as well as kinematics pattern under different conditions: barefoot, orthoses with shoe and insole with shoe.

Gait analysis is a motor skill studied by many investigators, since the bipedal plantigrade progression is a human ability that, in part, is learned (2).

This study intends to evaluate the gait progress of this child, the effects of foot orthoses' use, taking in consideration the disease progress, which is reported by the medical reports, and complementary exams done regularly, such as magnetic resonance, computerized axial tomography and others described later in the subject's characterization.

The effect of the therapies that the child attends, such as physiotherapy, should be considerate in the disease progress.

This description can be helpful to others and in the future a support for those with the same disease. It can contribute to the future description of a therapeutic plan according to the present results. According to Martin Bax (3) it is helpful to measure the movement of the young child with movement problems *in order to assess the effectiveness of the wide range of treatments offered to children.*

The study also intends to evaluate and analyse the child's gait, with the aim of sharing the information with other technicians involved in the case, and find solutions to improve the child gait performance, the foot orthoses' effectiveness and the need of adaptations.

It is also important to correlate the data collected from all the means of diagnostic and regular exams, to write the child's profile, and in the future use it as a predictor of progress, which can be a the challenge.

In addition, and since few cases were described all around the world, and with variability between them, this is an opportunity to start describing the disease biomechanically, sharing with the scientific community the development and progress of one case.

The specific goals of this study are:

- To evaluate the subject's spatio-temporal parameters and kinematic gait patterns under the following conditions: barefoot, orthoses with shoe and insole with shoe.
- To evaluate the subject's spatio-temporal parameters and kinematic gait patterns differences over time under the following conditions: barefoot, orthoses with shoe.
- To correlate the biomechanics data collected with the data collected from the complementary exams and the diseases characterization whenever possible.

The first reason is to provide a description of the subject. Gait is a sensitive measure of neuromuscular development or impairment, and a sound descriptive model is useful in defining normal versus abnormal gait, giving some clues as to the underlying cause of abnormality. Secondly, by following children through a treatment program it becomes possible to predict what is likely to happen in future cases (3).

2. Literature Review

2.1. Introduction

It is important to understand what *Cerebroretinal Microangiopathy with Calcifications and Cysts*, the subject's diagnosis, is and what characterizes it.

After that an overview about neural development and plasticity, and about visual impairment, will help to understand the child disease and its development.

Once that the focus of these case study is the child's gait development a literature review about this theme will be also presented.

Martin Bax in the foreword of Sutherland's book wrote that it is completely necessary *to have clear data on the normative gait of young children* (3). Normative gait is very important when investigators measure and describe a child with movement problems, since those results must be compared with the standardized for normal children (those who do not have movement problems or a pathological gait), in order to establish the problem's dimension and characterize it.

In the present study the normality description is crucial to understand the similarities and differences to the results obtained from data collected, and understand how the disease influences this child's gait performance.

The specific movement tasks change from individual to individual (4); that's why, in the present study, the specificity of the child's gait, and more specifically the detail of each joint movement involved in the gait cycle and the movement performance is observed.

Biomechanical analyses will be helpful to compare the subject with problems to the normal population and describe the changes observed (4). The information collected may lead to improve treatment planning, as well as recommendations for therapy (5).

The result of the analyses will allow the possibility to help this child to walk at his maximum of capability, adjusting techniques to stimulate independent walking until the best answer to his problem is found (4).

2.2. Cerebroretinal Microangiopathy with Calcifications and Cysts

Cerebroretinal microangiopathy with calcifications and cysts (CRMCC) is a rare disease, characterized by cerebral cysts and calcifications, with leukoencephalopathy and retinal vascular abnormalities (1). There is a lack of information about the number of cases because it is a disease with few cases related.

Tolmie, Browne, McGettrick and Stephenson (6) had already related cases of people suffering from this disease, like one of sibling girls with bilateral *Coats* reaction, retinal angiomas, intracranial calcifications, hair and nail defects.

Tolmie, Browne, McGettrick and Stephenson (6) refer that until then this set of characteristics was not described, and that probably it is an autosomal recessive gene defect, which is in agreement with authors such as Gong et al. (2001) (1).

The probability of occurrence of intrauterine growth retardation, renal abnormalities and reduced occipito-frontal circumference with CRMCC was also identified (6).

According to Gong et al. (2001) (1) it is still not identified the genetic cause of this disease but some mutations that are associated have already been identified.

The disease presents skeletal modifications firstly described by Sazgar et al. (2002) (1) (7).

Some studies refer to compromised longitudinal growth pre and postnatal (7), generalized osteopenia or early onset low turnover osteoporosis with fragility fractures (7) and metaphyseal abnormalities that may lead to limb deformities such as short femoral neck or *genua*

valga (1). It is still difficult to describe the progression of the described points once that there are few studies published (6).

There are other modifications related in the papers, such as hematologic modifications related by Revesz & Fletcher (1992); Goutieres et al. (1999); Sazgar et al. (2002); Crow et al. (2004) (1) (7).

One of the death causes of patients with CRMCC is gastrointestinal bleeding, another problematic associated to the disease (7) which can be associated with cirrhosis.

Neuropathological findings suggested that the primary abnormality behind the profound cerebral changes is an obliterative angiopathy of small vessels. The modifications under the retina result from vascular abnormalities that include angiomas and/or telangiectasias with lipids exudate and avascular periphery (1).

The name given to this disease *Cerebroretinal microangiopathy with calcifications and cysts* (CRMCC), results from all the characteristics until now identified (Linnankivi et al. (2006) (1)).

Crow et al. (2003) (7) refers that CRMCC is added to a disorder of movements in the brain and at extrapyramidal level.

Briggs, Abdel-Salam, Balicki, Baxter, Bertini, Bishop et al. (8) described the affection of the white matter of the brain.

Some cases have associated to them spasticity, dystonia, ataxia, and losses in the cognitive field (8). There are also related cases of potential anemia (8).

2.3. Neural development and plasticity

The lack of opportunities for children to move freely and explore the environment, which can occur in children with health problems who need to stay in the hospital for longer periods of time and children with visual impairment or low/high muscle tone, may delay the development of locomotor skills (9).

It is quite remarkable the interest shown by researchers about nervous system. Many reasons support that interest, one of which is about the practical application that neuroscience has, which can help on the treatment or rehabilitation of some diseases (10).

The brain, located in the head, is for human beings the largest when compared with the body size, having its surface divided into four lobes: frontal, parietal, occipital and temporal (10).

Frontal lobe is on the anterior region, parietal lobe is on the middle region, occipital lobe is on the posterior region and the temporal lobe is below the parietal lobe, covering the lower parts of it and the frontal lobes. In most of the studied brains it is not clearly delineated where each of the regions finishes (10).

Three regions of the cerebral cortex, named as motor cortex, have the responsibility of motor control: primary motor cortex, premotor cortex and supplementary motor area (10).

Primary motor cortex lies anterior to the central sulcus, opposite to the somatosensory cortex. Premotor cortex lies anterior to the primary motor cortex, extending from the dorsal surface of the brain and down laterally in the parietal lobe. The supplementary motor area also lies anterior to the primary motor cortex but on the medial surface of the brain (10).

The primary motor cortex's functions are mainly guiding intended movements to their targets, and the other two areas have important roles in planning those movements. Together those areas will control visually guided movement, coordinate postural adjustments that must accompany a movement, and are responsible for planning a complex physical movement (10).

Visually guided movements can be disrupted by lesions in the motor cortex, which can occur outside the primary motor area. The supplementary motor area may help adjust motor output to the requirements demanded and it is responsible for planning complex movements (10).

Complex movements are also controlled with the support of other components of motor systems, such as: basal ganglia, thalamus, and cerebellum (10).

The maturation process involved on the independent human locomotion is a very complex one (3), since there are many components such as: learning, emergence of cortical control, sensory integration and myelination; that contribute, all together, to the emergence of independent human locomotion (3). McGraw (1940) and Scrutton (1969) (11) wrote that learning and central nervous system maturation contribute (together) to the mature gait evolution.

The central nervous system is the most complex biological system on the planet (12). This system has been studied for several years and one of the great achievements is the knowledge about the capability that the central nervous systems has, which is the capacity for neurons to structurally and functionally adapt to reorganize neural circuits.

William James, the first one to describe the "plasticity" wrote in *Principles of Psychology* (1890) (12) *Plasticity, then, in the wide sense*

of the word, means the possession of a structure weak enough to yield to an influence, but strong enough not to yield all at once. Organic matter, especially nervous tissue, seems endowed with a very extraordinary degree of plasticity.

Neural plasticity can be defined as any change in neuron structure or function that is observed either directly from measures of individual neurons or inferred from measures taken across populations of neurons. This definition is not specific to any region of the CNS or any one experimental approach or assay (12). ... behavioral measures alone do not tell us exactly how the CNS is adapting during treatment (12).

Brain plasticity is the term used to describe the constant cellular and intracellular modification that occurs at brain's level and that results in changes in the neurologic function (13). This term is a result of recent decades' research in the neurosciences, which shows that the brain is not a static organ; instead it is a dynamic organ which can continuously adapt and change in terms of structure and function (13).

According to Byers (1941) (3) at 7 ½ months of conception all the neuronal cells are born, and Rabinowitz (1964) (3) adds that the brain is more mature at birth than are the other organs.

According to Moreel & Norton (1980) (3) myelination is important once it permits the use of less energy to nerves conduction, faster conduction and clear reduction in the space required. Myelin is like an electrical insulator that increases speed with a small nerve fiber, so it is not needed a big size nerve fiber because a large number can occupy a small space (3). If myelin is present, it is unnecessary a huge flow of sodium ions and the conduction still salutatory, from node of Ranvier to node of Ranvier (3).

When there is a lack of myelin, it all occurs more slowly and more energy is required, also fibers will have to increase in size and the central nervous system will need to occupy more space (Morel and Norton, 1980) (3).

According to Grillner (1975) (3) the timing of muscle activity and the intensity of its contractions are controlled by spinal centers and peripheral nerves. In an adult mature gait, peripheral receptors that are allocated in the skin, muscles and joints provide afferent stimuli that is automatically processed by spinal centers to modify muscle action in walking.

Neuronal plasticity allows the central nervous system to learn skills and remember information, also to reorganize networks in response to environmental stimulation as well as to recover from brain and spinal cord injuries (14).

Neuronal plasticity can be due to a prolonged period of overproduction and pruning of synapses in children and young adults (14).

Neuronal plasticity is enhanced in the brain development and is adaptative and positive to the individual, although in some ways it can be negative and the cause of neurological disorders (14).

When an injury occurs in the brain some mechanisms can occur to recover from it such as reorganization in the regions closer to the injury, recruitment of ipsilesional and contralesional areas, shifts in interhemispheric interactions, and shifts in bihemispheric connectivity (15).

The acquisition of brain recovery is due to, and can be confirmed by, the link between basic science, medicine, imaging, and rehabilitative professional expertise (15).

That is why it is important to understand the mechanisms responsible for brain plasticity and how they could be influenced in order to improve positive results in the brain's damaged areas (14).

Nowadays most of the investigation in the neural plasticity area is done with rats, since they have highly evolved motor systems and extensive repertoire of motor abilities (12).

Studies of motor control plasticity, after stroke, have shown that recovery and compensation, at a neurophysiological level, occurred, calling recovery to the restoration of motor function within an area of motor cortex that was initially lost after injury, and calling compensation when areas of motor cortex adapt themselves to take on motor functions lost after injury (12).

Activity dependent of neuronal plasticity appears to play an important role in the evolution of clinical signs of motor dysfunction in children with cerebral palsy (14) and probably other dysfunctions not studied yet.

Plasticity, derived from the Greek word 'plaistikos' meaning 'to form' refers to the brain's ability to learn, remember and forget as well as its capacity to reorganize and recover from injury (Buonomano & Merzenich, 1998) (16).

It is clear for science that the central nervous system has the capability to adapt structurally and functionally in response to experience; those are the two categories of neural plasticity (12).

Neural circuits that control the trained movements reorganize themselves by adding synapses, which results in an expansion in the amount of cortex involved in movements control (12).

The brain is incredibly interconnected and this interconnectivity plays an important role in supporting functional reorganization after an injury or disease (12).

It is important to notice that neuroplasticity can occur through processes of restoration, recruitment, retraining (12).

Restoration is about the ability that residual brain areas have to restore abilities after a brain injury or a disease even being affected by that particular problem (12).

Recruitment is about the motor areas that have the capacity to contribute to the lost motor function. These areas are asked to help with the performance of the impaired motor behavior but that doesn't mean that they are acquiring a new function (12).

Retraining is observed when areas of motor cortex adapt or take additional functions to support functional improvement, more than restoration and recruitment, retraining means that those motor areas will develop for those tasks from now on (12).

Functional imaging and transcranial magnetic stimulation studies showed that after neurologic injury, brain "recruits" areas, usually distant and sometimes contralateral areas to cover the function of the damaged area, and this could happen in hours or days (13).

Also (13), recent studies have shown that brain plasticity is the result of factors such as: environmental interaction, pharmacological agents, trophic factors and environmental enrichment.

2.3. Visual Impairment

Vision provides important information for self-motion in the environment and is of high importance in motor control (17).

In tasks related to motor control, two kinds of vision assume an important role. Focal vision, mediated by visual information from the central retinal field, is responsible for detecting physical information of the objects displaced over the environment, and ambient vision, mediated by peripheral vision, is responsible for detecting the characteristics of those objects (17).

The importance of visual system is its ability to extract from the images collected by the eyes, information about the shape, location, and movement of objects in the environment (10). The visual system provides information about the environment from a distance, it is important during locomotion in the maintenance of stability and for route planning (18) (19) (20).

Vision is dominant in the control of the direct goal of locomotion (21). The information collected allows orientation towards a goal, adjusting heading direction, avoiding collisions with objects, avoiding obstacles and accommodating different surfaces.

Visual perception of self-motion, limb position and limb movement is important in order to adjust foot clearance or foot placement, as well as to regulate walking speed (18).

The information that vision provides is due to high speed of propagation of light waves, this allows the interpretation of information and the opportunity to take anticipatory actions before any situation occurs. Those actions are classified as strategies and include: *(a) selection of alternate foot placement by modulating step length and width; (b)*

increased ground clearance to avoid hitting an obstacle above ground; (c) changing the direction of locomotion (steering) when the obstacles cannot be stepped over or under, and (d) stopping (20).

Investigators found that global motion is affected by a number of genetic and acquired developmental disorders, such as visual deprivation, as well as neurodevelopmental disorders and its relation with visual deprivation (22). This theme is becoming interesting to investigators.

In a normal vision photoreceptors area arrayed along the back part of the eyeball, in the layer of receptor cells and neurons, named retina (10).

The light enters into the eye, through the transparent cornea, passing through the pupil and the lens, falling on the retina. The light, which enters in the eye through the transparent cornea, passes through the pupil and the lens falling on the retina. The pupil, which is in the middle of the iris, adjusts its own size according to the intensity of light received controlling the amount of light that will reach the retina. Light is focused on the retina by the lens, whose shape can be changed by intrinsic muscles of the eye. The optic nerve, cranial nerve II, carries visual information to the brain. Fovea is a specialized region of the retina that is adapted for acute vision, having a dense array of photoreceptors; it is also a special region of retina where the light falls from objects the animal looks at directly (10).

The travel that visual information does starts when information about light enters the brain via the axons that form the optic nerve, travelling to lateral geniculate nuclei, a pair of nuclei in the posterior part of thalamus. Then information travels to the primary visual cortex (also known as the visual cortex, area VI) and to the striate cortex (the first cortical area developed to process visual information) (10) (23).

The information is transmitted to primary visual cortex via three distinct sub-pathways: the magnocellular (M), the parvocellular (P), and the koniocellular (K) (23). These cells differ on their location: M are the lower two and large cell bodies, P are the upper four and smaller cell bodies, and K are interlaminar to each of the other six. All those cells differ in their physiology as well as in their anatomy (23).

M cells are more susceptible of developmental damage since they are bigger and at the same time less, so if something happens to some of them, rapidly an effect will be seen (23).

Cortical projections from the visual pathways proceed in a hierarchical organization, from the lower to the upper cortical areas (23).

Visual signals are (also) sent to the superior *colliculi*, a pair of domelike structures in the midbrain, and to the *pretectal* area that lies just anterior to the *colliculi* in the midbrain. The information sent to the *colliculi* helps controlling movements of the eyes to new targets and the information sent to *pretectum* forms part of a circuit for reflex control of the pupils (10).

Beyond the areas until now described, more areas from visual cortex are important in the visual information process; those areas are located on the middle temporal region, and more areas can contribute to the processing of visual information (10).

There is a critical period for the development of binocular vision but also of the entire cortical visual system, during which visual input must be present in order for it to develop normally. Deprivation can have more or less effects depending on the period in which it occurs. If occurring during the critical period of the system development, it will have profound and permanent disturbs (10).

It seems that children's gait is more susceptible to the removal of vision than the adults' gait (19) since children with ages between 2 and 5 are more affected by any disturbance in the visual system (21).

Despite all the locomotion adaptations identified, Houwen et al. (2007) (24) did not identify any difference in locomotion skills between two children with 9 years-old, one with visual impairment and the other one sighted.

According to Sutherland (3), in normal walking, visual stimuli have less importance than kinesthetic stimuli when we are talking about limb position, although it is important for the person to have information about the space and the objects that are closer. However, others have different opinion and stated that vestibular and proprioceptive information are not enough to compensate the loss of vision (21) (25).

All the types of information are important, but sensory input that becomes from the vestibular system derived from sway-related linear and angular accelerations of the head may be more important, as it contributes to balance and walking. Although it seems to be like so, only when the others were lost this one takes advantage (Begbie, 1967; Nasher et al.; 1982) (3).

It is necessary to have an efficient conduction system to link peripheral receptors with the central nervous system, where the motor commands took place. Also it is necessary to have an efficient cortical control, efficient conduction system and interneuron connection to achieve mature gait (3).

Vision impairment results in different locomotion adaptations such as decrease in walking speed (smaller steps are taken), decrease in step length, increase in the foot placement error, deviations from a straight path, more backward leaning position of the trunk and changes in foot

positioning (the initial contact is made with the entire plantar surface of the foot instead of a clear heel strike), less ankle plantar flexion following heel strike (18) (19) (24) as well as a prolonged duration of double support phase (a way to recover balance and return to a controlled phase). Hallemans, Beccu, Van Loock, Ortibus, Truijen, Aerts (21) add that stance phase is also prolonged, giving more time for the foot to be in contact with the ground and probe the ground. Changes in cadence, swing limb trajectory, foot elevation are also described.

Adding more gait modifications to those above, some other kinematic modifications are referred by Hallemans and colleagues (25), just like decreased hip adduction in stance, increased knee flexion at foot contact, decreased foot roll-over at loading response and decreased ankle plantar flexion at push-off.

It is also important to pay attention to the reduced range of motion (ROM) of pelvis, which can be a strategy to reduce the degrees of freedom of movement and have more control over them (25).

In Hallemans and her colleagues' (18) opinion, typical adaptations are: smaller stride length and a more plantar foot contact, as result of a strategy to face visual deprivation and maintain stability. Those adaptations can be interpreted as a safer walking strategy (19).

Visual system is also a theme under constant study and one of the aims of those studies is the system's plasticity (22).

Development of independent walking can be a more challenging task for a child with visual impairment than for others with normal vision (24).

2.4. Gait cycle

Gait cycle is about *the movements and events that occur between successive footsteps of the same foot (3)*, which is the same of saying that starts with a *heel-strike (HS)*, *continues through stance and swing phases, and ends with heel-strike of the same foot (3)* or the period from the foot's initial contact until the next foot initial contact (26).

There is a close relation between gait cycle events and joint angles. Since toe-off is expected to occur with the maximum plantar flexion and hip abduction, opposite toe-off is expected to occur at 50% of gait cycle, almost happening at the same time of maximum hip extension, maximum external rotation of the pelvis, maximum internal rotation of the hip and maximum hip adduction (3).

It is possible and usual to divide gait cycle in two phases: swing and stance, as the result of foot contact with the floor or not (26). During a normal gait cycle, it is possible to observe that stance phase takes approximately 60% of the cycle and swing phase about 40% (26).

Stance phase could either be divided in single support and double support, according to the contact of only one foot or both feet (26) with the floor. Stance phase can be divided in different events such as heel strike, foot flat, mid stance, heel off and toe-off (26).

Swing phase could also be divided in three different phases such as early swing, mid swing and late swing (26).

Because of all those particularities involved in gait cycle, those events must be studied singly, one by one, but also all together, since the result (gait) is the parts' sum (2).

2.4.1. Spatial Reference System

The spatial reference system can be relative or absolute. If it is a relative system, it requires all the coordinates to have to be reported relatively to an anatomical coordinate system that changes from segment to segment. In case of an absolute system, it means that the coordinates are referred to an external spatial reference system (4).

Gait can be described over the three different planes: sagittal plane, frontal plane and transverse plane (4) (27).

2.4.2. Gait Cycle Spatio-Temporal Parameters

According to literature review, there are important parameters to study which help with gait description, although there is not a consensus about which parameters are more important to describe.

If the aim is to describe a gait cycle, it is important to measure parameters such as: step length, stride length, opposite toe-off, toe-off, single-limb stance duration, cycle time, cadence and walking velocity (3) (11), those parameters are a good indicator of gait maturation or identification of gait problems (3).

Although in Murray et al. (1964, 1970) and Rigas (1984) (26) opinion, to describe the spatial parameters of a subject's gait, it is necessary to consider the following spatial parameters: step length, stride length, foot angle and base width.

If the aim is to evaluate gait maturity, five parameters have to be described: single limb stance, walking velocity, cadence, step length, and the ratio of pelvic span to ankle spread (11).

When analyzing gait, to describe temporal parameters, it is also important to consider parameters such as cadence and average velocity (26), being extremely important to record the time of consecutive heel strike and toe-off in order to obtain step and stride time. Other interesting parameters like single support time and double support time can be obtained and would also be important to study (26).

In summary, some of the spatial and temporal parameters referred are:

- Step length is the distance between initial contact by different foot (right/left or left/right) (26)
- Stride length is the distance between two consecutive initial contacts by the same foot (right/right or left/left) (26)
- Stride width is the medial-lateral distance between consecutive heel strike (19)
- Step time is the time between two consecutive heel strikes. It represents 50% of the stride phase time if the person walks with perfect symmetry between the left and the right sides (26)
- Double limb support is the time over which the body is supported by both legs. It takes two periods during a gait cycle, each of them lasting 10% of gait cycle (26)
- Swing time is the time that leg takes to swing through, while the body is in single support on the other leg. It takes the same time of single support (26)
- Stance time is the time over which the body is supported by one single leg (26)

- Single support is the time over which the body is supported only by one leg, which is approximately 40% of gait cycle (26)
- Cycle time is the time necessary for two consecutive heel strikes with the same foot and includes stance and swing phases (19)

Because there are no known studies about those parameters applied to Portuguese children and few studies in the international literature, Sutherland and colleagues' study (3) was used as reference. So, in their study by age, it is presented the mean time / distance parameters for 5-year-old normal subjects.

Time/Distance Parameters (3)	
Opp. Toe-off (% cycle)	13
Opp. Foot strike (% cycle)	50
Single Stance (% cycle)	37
Toe-off (% cycle)	63
Step length (cm)	42
Stride length (cm)	84
Cycle Time (secs.)	0.77
Cadence (steps/min.)	153
Walking velocity (cm/sec.)	108
(m/min.)	64.8

Table 1 – Mean time/distance parameters for 5-year-old normal subjects

2.4.3. Gait Cycle Kinematics

The output of the movement is what we see (4) and can be described by a large number of kinematic variables, which are involved in the description of the movement, independently of forces that cause that movement. That includes linear and angular displacements, velocities and accelerations (4).

The correct functioning of the joints movement patterns involved in the movement allows a smooth and energy-efficient progression of the body, meaning that the relation of lower limb joints (ankle, knee, hip) is critical, and any abnormality in co-ordination could have a negative influence in gait proficiency (26).

Having an accurate model of the human body with correct anthropometric variables, it is possible to develop a reliable link-segment model (4) .

The displacement data is taken from any anatomical landmark: center of gravity of body segments, centers of rotation of joints, extremes of limb segments, or key anatomical prominences (3).

It is impossible to develop a biomechanical model without data regarding masses of limb segments, location of mass centers, segment lengths, centers of rotation, angles of pull muscles, mass and cross-sectional area of muscles, moments of inertia, and so on, it is necessary to complement one kind of data with others (4).

When describing joint motion patterns, during gait usually (26), the described movements are: ankle plantar-dorsiflexion, foot rotation, knee flexion-extension, knee valgus-varus, knee rotation, hip flexion-extension, hip adduction-abduction, hip rotation, pelvis tilt, pelvis

obliquity and pelvic rotation. Those movements correspond to joints described over the three different planes.

2.4.3.1. Gait Cycle Description over Sagittal Plane

In the sagittal plane, the described movements are plantar-flexion and dorsiflexion for the ankle, and extension flexion for knee and hip. In this plane, dorsiflexion is positive and plantar flexion negative, knee extension is negative and knee flexion is positive, hip extension negative and hip flexion positive, pelvis anterior flexion is positive and posterior flexion is negative (4) (27).

2.4.3.1.1. Ankle

Foot moves as a whole through the tibia, and this movement is considered ankle motion, and is usually reported as a movement of foot and ankle as a whole (26).

Plantar flexion and dorsiflexion movements are of great importance because they allow shock absorption at heel strike and during the stance phase, being also of great importance to the propulsive stage right before the instant when toe leaves the floor ("push off" movement). The range of ankle motion during gait cycle can vary between 20° and 40° for normal adult gait (26).

Ankle joint is also important during swing phase once it allows foot clearance, which in a pathological situation could be described as drop foot (26).

The ankle movement over this plane in the mature gait (3) is represented by a curve with two peaks and two valleys.

During gait, ankle has four phases of motion (11) (26).

During the first phase of motion it is possible to observe (in normal situations) that, at initial contact, ankle is in a neutral position, turning to plantar flexion in a movement between 3° and 5° (26) in a maximum of 6.8° (27) until foot flat. Sometimes that is called foot pivoting about the heel or calcaneus (26).

When heel contact with the floor occurs there is a small dorsiflexion to lower the foot to the ground. Following there is a large increase in plantar flexor moment reaching a peak at about 50% of stride to help ankle to rapidly plantarflex and achieve an upward and forward “push-off” of the lower limb as the subject starts swing at toe-off (4).

The first valley result of ankle plantar flexion and occurs at more or less 8% of the cycle (11). At 1 year-old that is not observed, once at this age child strike flat-footed, only after 1 ½ years-old it starts to be observed (3).

During this phase, plantar flexion is controlled by dorsi flexor muscles acting eccentrically. The role of these muscles is to help in shock absorption and aid weight acceptance to the lower limb (26).

On the second phase of ankle motion, with the foot flat, ankle begins to dorsiflex progressively (26) (27). During this movement, the foot remains stationary while tibia becomes the moving segment, moving over the ankle joint, and dorsiflexion reaches a maximum of 10° (26).

Ankle dorsiflexion proceeds through single limb stance, peaking at around 40% of the cycle (11) (3). During this phase, in order to control the movement of tibia forward, plantar flexor muscles act eccentrically (26).

After this movement, a degree decrease until the opposite foot-strike, happens at 50% of the gait cycle (11), that happens because the direction of motion reverses, ankle joint acceleration begins and dorsiflexion slowly diminishes (3).

On the third phase, the heel begins to lift at the beginning of double support, causing the prompt plantar flexion movement, that will range an average value of 20° at the end of the stance phase, at toe-off, and the pivot point now is under the metatarsal heads (26). This is a propulsive phase, associated with increase of angular velocity in the ankle, which can achieve $250^{\circ}/s$ of angular velocity for normal adult gait. Plantar flexor muscles will contract concentrically to push the foot into plantar flexion and propelling the body forward (26).

After rapid plantar flexion, which reaches a maximum at toe-off (11) (3) dorsiflexion begins.

That is the beginning of phase four, swing phase, when a rapid ankle dorsiflexion occurs, achieving an angular velocity of $150^{\circ}/s$, to allow the foot clearance from the floor. At mid swing, a neutral position is achieved by ankle, 0° , maintaining it during the swing phase until the next heel strike. It is possible to observe during this phase a 3° to 5° of dorsiflexion (26).

Ankle dorsiflexors contract concentrically in order to provide foot clearance from the floor and prepare the next foot strike (26).

Dorsiflexion is observed until the beginning of plantar flexion during late swing phase (11) and it seems that the action of the ankle plantar flexors, during this phase is very passive without EMG activity (3).

2.4.3.1.2. Knee

In the sagittal plane, the knee movements are flexion and extension with a range of motion that varies between 0° and 70° for normal and mature gait cycle (26). Even so, the instant when peak flexion occurs can vary due to: differences in walking speed, subject's individuality and the landmarks selected to define limb segments (26).

Knee's flexion/extension patterns are divided into five phases (26).

At heel strike, or initial contact of foot with the floor, the knee should be flexed, although people's knee posture can vary between slightly hyperextension (-2°) to flexion (10°), with a mean value of 5° (26). During each gait cycle knee flexes twice (11).

Phase one of knee's movement occurs after initial contact, when a flexion of 20° is observed and flexion takes maximum weight-bearing loading. This flexion movement occurs in order to absorb the load at a rate of 150° to $200^{\circ}/s$ (26). The knee extensors are active at 8 to 25% of stride, to control knee flexion as the limb accepts weight (absorption phase), producing a gradual elevation of the body's center of mass and reducing the energy required for walking (Inman et al., 1981) (3) (2). A flexor pattern as a by-product of the *gastrocnemius*'s contribution to the ankle plantar flexor moment will be observed (4). At the same time, ankle's plantar flexion occurs and both (ankle and knee) act together to absorb shock during the loading phase of lower limb.

Knee extensors act eccentrically (3) and knee flexion velocity slows as the knee reaches initial flexion peak (26) .

Two children groups were studied and they show a 10° increase in the knee's flexion after foot-strike, followed by a decrease in flexion until

35% to 40% of the gait cycle when the knee flexion angle is approximately the same measured at the heel-strike (11).

After that the extension movement occurs, at a rate between 80°/s and 100°/s, almost achieving full extension. This is a controlled movement of body over the stance limb (26). As the body passes the leg, the knee extension and the foot plantar flexion allow the heel to rise and this result in a relative leg elongation (2).

The third phase is characterized by a coincidence of heel lift (heel begins to lift at 50% of the gait cycle) with a new knee flexion. This occurs during lower limb propulsive phase, when knee presents a rapid flexion preparing itself to swing phase (sometimes this is called pre-swing phase) (26).

The fourth phase correspond to toe-off, which happens at the same time of knee's flexion, this flexion has approximately 40° at a rate of about 300 to 350°/s, being possible to observe the flexion wave occurring right before toe-off, in preparation to the limb advance (3).

Right before and after toe-off, a small knee extensor moment acts to limit the amount of flexion in late stance and early swing (absorption phase) (4). The knee velocity then slows until when it achieves maximum flexion, occurring at the same time of ankle dorsiflexion, promoting the toe to clear the floor. Between initial to mid swing the knee is still flexing until 65° to 70° (26).

The last phase occurs during the later swing when the knee undergoes a rapid extension at about 400°/s to 450°/s and this movement will prepare second heel strike (26).

The final burst of flexor activity right before heel contact has the aim of decelerate the swinging leg prior to heel contact (4).

2.4.3.1.3. Hip

Hip joint angle is defined as the angle between pelvis and thigh segment (26).

During the movement's first phase, after heel strike, hip extends as long as the body moves over the limb, this occurs at a rate of $150^{\circ}/s$ and hip achieves maximum extension right after opposite heel strike (26).

After maximum hip extension, a new phase begins. The change between extension and flexion occurs at the same time that opposite foot-strike takes place (3); at this time (opposite foot-strike) the hip flexion is in average about 40° (11).

Weight is transferred to the forward limb and the trailing limb begins to flex at the hip (pre-swing period). At 60% of gait cycle the foot leaves the floor and hip flexes quickly at a rate of $200^{\circ}/s$, this will help the limb's forward progression to take a step. Hip reaches its maximum flexion right before heel strike (26). Hip's flexion starts during the second double support (3).

Third phase is characterized by a hip's small extension movement becoming less flexed. That movement occurs in order to pre-positioning the foot right before heel strike (26).

During gait cycle the leg flexes forward and the hip joint goes forward extending right after, until push off (26).

Flexion peak occurs at 85% of the gait cycle (3), reaching a maximum of 62° during early swing phase and diminishing approximately 5° at foot-strike (3).

The hip has an extensor moment for the first half of stance that is followed by a flexor moment in the later half (4).

In the first half the extensors have to stabilize the posture of trunk preventing it from flexing forward in result of the large posterior reaction force at the hip; this also helps the knee's extensors preventing collapse of the knee's joint and contributes to forward propulsion "push from behind" (4).

During the second half of stance, the flexor moment is needed to stabilize the trunk and preventing it from flexing backward, and also, in the last phase of stance and early swing (50-75% of stride), to achieve a "pull-off" of the thigh into swing (4).

2.4.3.1.4. Pelvis

Over this plane pelvis movements are controlled by gravity, inertia, and the action of the hip flexors: *iliopsoas*, *rectus femoris*, *sartorius* and *tensor fascia femoris*; and extensor muscles: *gluteus maximums* and the *hamstrings* (3).

The pelvic movements, on this plane, consist in small variations in the degree of anterior pelvis tilt, which is inclined downwards anteriorly (3). Pelvic tilt is variable between 6° and 8° (2).

It is desirable to obtain two peaks and two valleys. The valleys take place when toe-off and opposite toe-off take place, and peaks occur at late single stance and mid-to-late swing phase (3).

Anterior pelvis tilt diminishes as the limb is loaded during initial double support. It then increases as the body's center of mass moves forward over the supporting foot, decreases again during push-off (between

40% and 50% of the cycle) to a low point at toe-off and finally increases until following foot-strike (3).

2.4.3.2. Gait Cycle Description over Frontal Plane

In the frontal plane, the movements described for the ankle are inversion and eversion, for knee and hip are abduction and adduction, and for pelvis is obliquity. In this plane, eversion is positive and inversion is negative (4) (27), abduction is positive and adduction negative, pelvis obliquity is positive or negative depending on the movement up or down.

2.4.3.2.1. Ankle

Ankle's inversion and eversion movements are described by foot as a whole (26).

At heel strike, foot lands in inversion, moving to eversion during loading phase (26).

Then a quickly inversion takes place just prior to 50% of gait cycle. A range of motion of 7° is observed (26) or 7.3° (27).

2.4.3.2.2. Knee

Similar to what happens with the hip, which prevents the pelvis from dropping, the knee in response to the weight-bearing and to the large gravitational load, tries to invert but the passive loading of the medial condyles and unloading of the lateral condyles creates an internal abductor moment (4).

In the frontal plane, knee presents not only variance between individuals, even though standard deviation between them is very low, but also effect of cross talk from other planes (26).

Stance phase presents more reliable measurement than swing phase. During stance phase, for normal individuals, there will be little movement besides *a slight deformation during loading and the opposite deformation during terminal stance* (26). This deformation is based on anatomical alignment of the knee (adduction/abduction or valgus/varus) (26).

During stance phase it is not expected to observe more than 4° of movement (26).

During swing phase more movement can be recorded, but *it is unclear if this movement is real and due to the laxity of the joint or if it is an artifact due to the change in the orientation of the segment coordinate systems* (26). It is possible to observe 10° of movement (26).

2.4.3.2.3 Hip

Three phases are identified during a gait cycle (26).

Phase one is the period when heel strike occurs and hip is in a slightly abducted position (26) or in neutral position (3), after moving quickly into adduction (26) (3) at the same time that limb is being loaded and the body supported. This adduction movement is due to the dropping down of pelvis during contralateral limb (26).

The second phase is an abduction movement. During this abduction movement, pelvis levels go out and the body progress over the stance limb. Hip reaches the maximum abduction shortly after toe-off, as the pelvis drops down, and limb is now on swing phase (26) and declining

rapidly to a plateau at 30% of the cycle until opposite foot-strike at 50% (3).

The third phase is when pelvis levels are off and the limb swings through mid to late swing phase. The range of motion is about 15° , with equal amount of adduction and abduction (this movement is closer to the one that describes pelvis on the frontal plane) (26).

It is expected to observe the abductor muscles acting eccentrically in order to decelerate hip adduction during initial double support and early single support. Then it will be necessary to produce eccentric action of hip adductors to stabilize the hip, thus decelerating hip abduction, since the force produced to move forward will pass from the lower limb's medial side to the lower limb's lateral side (3).

Maturity in this movement is achieved at 2 ½ years old, and it is not expected to observe any change after that age (3).

Immature age groups present less adduction in stance phase and a lower overall dynamic range of motion and the younger children walk with a wide base that influences their lower adduction movements (3).

In the frontal plane, at stance, there is a strong abductor pattern which prevents the pelvis from dropping against the forces of gravity that are acting about 10 cm medial of the stance hip (4).

2.4.3.2.4. Pelvis

In the frontal plane, pelvis movement is known as obliquity and occurs with the purpose of allowing shock absorption and limb length adjustments (26).

In the frontal plane during early stance phase, the contralateral side of pelvis drops downward. The peak of pelvic obliquity occurs right after opposite toe-off, which corresponds to early stance phase on the weight-bearing limb (26). This pelvic movement is important because it gives pelvis the opportunity to drop on the side of the non-weight-bearing, or swinging leg (2). Anterior superior iliac spine rises during weight-bearing to peak elevation when opposite toe-off occurs (3).

After that it drops, reaching a low point halfway through single stance, followed by a smaller peak elevation when opposite foot-strike occurs (3).

After foot-strike, the ipsilateral pelvis drops to its lowest level until toe-off (3).

The peaks are correlated with opposite toe-off and opposite foot-strike for every age except for those with 1 year old, for whom the first peak precedes opposite toe-off.

The dynamic range of motion varies across the age-groups between 4.2° and 9.4° , with a mean of 7.7° (SD 1.8°) (3).

The effect of pelvis motion in the frontal plane is the decrease of center of gravity amount, permitting the torso movement over the weight-bearing leg (2). The non-nullity is important to promote the movement (2).

2.4.3.3. Gait Cycle Description over Transverse Plane

In the transverse plane the movements described are external and internal rotation. External rotation is negative and internal rotation is positive (4) (27).

2.4.3.3.1. Ankle

Foot strike normally occurs with an 8° external rotation of the foot (3) continuing during early and mid-stance phase (11).

The external rotation diminishes slightly during initial double support because weight is uniformly distributed over all foot surface (3). After, foot rotates internally until toe-off (11).

The average dynamic range of foot rotation in this plane is about 10° for all the groups of children evaluated by Sutherland and his colleagues (3), the same author did not find significant differences (11) when comparing a group with two year-olds with one with seven year-olds.

2.4.3.3.2. Knee

In the transverse plane, the knee movement is dominated by the motion of tibia rotating through the femur during swing and stance phases (26).

According to Andriacchi (2005) (26) *this motion externally rotates the tibia through swing in to stance in order for the tibia and foot to be in the correct alignment at initial contact.*

According to Inman et al. (1981) (3) the relation of tibia with subtalar joint produces foot pronation in the early stance.

In the transverse plane, the tibial rotation is internal when loading phase of initial double-limb support takes place (3).

Following, the tibia rotates externally until toe-off, turning to internal rotation (3). At foot-strike tibia is in slight external rotation (11).

In total, between the maximum internal rotation to the maximum external rotation, there is a gap of 20° (11).

During foot-strike, two-year-old children have highest external rotation when compared with seven-year-old children (11).

2.4.3.3.3. Hip

In the transverse plane, the hip movement is the result of femur and pelvis movement, which can be described as internal and external rotation (26).

At heel strike, the hip is approximately at 10° of external rotation, but as the foot approaches the floor and the knee flexes, the body starts to move over the stance limb and an internal rotation occurs at about 5°, at the same time pelvis rotates forward on the swing side (26) (3) (11).

The internal rotation peak happens when opposite heel strike occurs, returning to external rotation (11). The hip shows a quick movement back to external rotation at late swing phase (26) (11) (3).

The dynamic range of motion across all children groups is about 15°, and the changes after 2 ½ year-olds are small (3).

According to Sutherland (3) passive external rotation exceeds internal rotation at birth, decreasing (in terms of range) with age, what can be explained by acetabular retroversion or soft tissues constraints which limit internal rotation in the youngest groups.

The major transverse activity at the hip, is during the first half of stance when the external rotators of the stance limb act to decelerate the horizontal rotation of the pelvis and the trunk over the stance limb, then

in the second half, the internal rotators are active to stabilize the forward rotation of the pelvis and swing limb (4).

2.4.3.3.4. Pelvis

Pelvis rotation occurs on a vertical axis changing between left and right. This rotation is usually about 4° on each side. The peak of internal rotation occurs at foot strike and the maximal external rotation peak occurs at opposite foot strike (26).

The importance of this rotation is that it lengthens the limb by increasing the step length and preventing excessive drop of the center of mass of the whole body, and in consequence, the walking pattern is more efficient (26).

Pelvic rotation also smooths the vertical excursion of the center of mass and reduces the impact of foot strike (26).

Pelvic rotation occurs alternately on each weight-bearing hip joint (2).

As the swinging limb is advanced, pelvis rotates internally, and the peak of internal rotation occurs slightly after foot-strike. (3). Pelvic rotation decreases in a faster way when heel strike occurs and is reversed as full weight is placed upon the foot (2).

With load acceptance, the pelvis begins a counter-rotation (external) through the weight-bearing femur when toe-off takes place, continuing to opposite foot-strike when internal rotation begins again (3).

This rotation, even with individual variation, can vary (2) between 6° and 8° (those are mature gait's references) and helps decreasing the amount of fall of the center of gravity with each step (2).

It seems that there is no significant difference between the two-year-olds and seven-year-olds curves (11), pelvic rotation, is slightly across the child's age.

2.5. Considerations about children gait cycle

The walking patterns of youngest children differ from adults' or mature patterns, that is why it is necessary to measure walking pattern at childhood and at different ages (3), since gait maturation takes a short period of time to occur (11).

Some known characteristics of gait changes with age and maturity. Cadence decreases while walking velocity and step length increase, since limb length increases and people achieve greater limb stability (11).

There is not a consensus about the age when children achieve mature gait, but according to Sutherland and focusing on the criteria above, mature gait is achieved at three years old (11). However, according to McGraw (1940), Scrutton (1969), Statam & Murray (1971) (11) there is a consensus on the fact that the development of walking skills is completed at five years old. Nevertheless, Burnett & Johnson (1971), Hennessy, Simon & Reed (1977), Sutherland (1966) (11) have found evidence of earlier gait maturation.

Inman (2) reinforces that the first faltering steps will give place to stable and precise steps, and Popova (1935) (2) suggests that only when a child reaches the age of 7 to 9 years old, does have a walking pattern closer to the adult.

Sutherland and his colleagues (3) reinforced that it is very important for a child to walk at free speed, and that many changes occur in the time/distance parameters from age 1 to 7 years-old, some of them related to changes in size of body segments, others probably caused by maturation of the motor control system.

Inman (2) adds that when a subject is able to choose the walking speed, he will always choose the velocity that has less energy expenditure for him, so the most comfortable.

Sutherland and his colleagues in their study about children gait development refer important points about gait development. With age, duration of single limb stance tends to maintain, without a huge increment, at the same time walking velocity does not seem to decrease because of that. Cadence tends to decrease with age, a fewer number of steps per minute. Step length increases rapidly until one-half/two years old, starting to increase slowly right after that. The ratio of pelvic span to ankle spread rises rapidly until one-half/two years old, then increases slowly until three years old remaining equal until the age of seven (11).

Also, in the sagittal plane, the children's angular rotations from two years old on are very similar to those of mature/normal adults (11). Also the reciprocal arm-swing and heel-strike are present in most of the children by the age of eighteen months (3).

Heel-strike, knee flexion wave and reciprocal arm-swing have been used as gait maturation indicators, but this evidence is not clear, although the absence of them is a huge indicator of pathological gait (11).

Sutherland and his colleagues (3) add that: *Height and leg length are directly related to step length. Age is directly related to leg length and step length; however because of a change in velocity of growth, the slope declines, beginning at 2½ years for leg length and at 4 years for step length.*

They also add that *normal children walk symmetrically: right and left step length are equal, or nearly so, and gait events also are equal, or*

nearly so. Walking velocity increases with age and in spite of decreasing cadence; this increase is due to increasing stride length. Cycle time bears an inverse relationship to cadence. Cadence decreases and cycle time increases rapidly between 1 and 2 years. Duration of single-limb stance and time of opposite swing are, by definition, equal.

And that: With increasing maturity single-limb stance, step length and walking velocity increase and cadence, initial double support and second double support diminish.

Also: By 4 years the inter-relationships between time/distance parameters are fixed, though stride length and walking velocity continue to increase with increasing leg length.

The greatest pelvis tilt is observed in the 1 and 1 ½ year-olds (3).

In terms of motion, the knee flexion is mature when we talk about knee-flexion wave in stance phase, at 4 years-old. The difference in the dynamic range of motion in a children group is from about 9° less in those who have 1, 1 ½ and 2 years old to those who are older (3), increasing 8° to 9° after heel-strike in the adults (11).

2.6. Foot Orthoses

The pathological position of the foot may lead to an increase in the load applied to the foot's ligaments, muscles and tendons, which can result in chronic injuries, also it will have influence in the propulsive movement of foot during gait cycle (Dennis et al, 1985; McCulloh et al, 1993) (28).

In order to improve posture in the foot and ankle, foot orthoses are prescribed. The use of foot orthoses can also be a way to release malalignements in the leg (29).

The use of foot orthoses should be preceded by a physical evaluation and biomechanical assessment of the foot in addition to the trunk and lower limb (30).

Foot orthoses are shaped or moulded inserts for the shoe and intend to hold the foot in position, change the foot's position, or either, change the foot's range of motion of whole foot or between different foot segments (26) presenting different shapes and forms, as well as being built in different materials.

The orthotic effects can depend not only of their type (posting, molding, or a combination) but also on their surface texture to stimulate the sensory feedback (proprioceptive orthoses) (31).

Most of the orthoses aim to affect a joint directly, although many orthoses have additional effects under distal or proximal joints, such as knee, hip and pelvis (26). However not every author recognizes the effects of orthoses' use as important, for example, Nester and his colleagues referred in their study that the effects of the orthoses on knee, hip and pelvis kinematics in general were minimal (32).

Following their idea, Nester and his colleagues referred that looking to the minimal effects that orthoses had on proximal joints to the foot, it seems that orthoses may have additional effects on the passive and active soft tissues of the lower limb, which contributes to the success of orthoses use (32).

Nicolopoulos, Scott, Giannoudis (29) advertise for the injuries on the lower limb that can be caused by the overuse of orthoses.

Orthoses effect might occur through the effect caused over proprioception mechanism involved in muscle function regulation (33).

Certainly, the success of a functional orthotic is dependent on clinical examination of the foot pathology, the biomechanical examination, the fabrication technique, the casting technique and the education of the patient (29).

Foot orthoses affect most the stance phase of the gait cycle, which is about 60% of gait cycle (29).

Functional foot orthoses are used either to correct and compensate biomechanical abnormalities at points throughout the weight-bearing chain (29). It is important, in the orthoses prescription, to pay attention to the relation between foot and the ground and also between foot and proximal segments, this requires legs' static and dynamic measurement (29).

The use of functional foot orthoses has been a success in lower limb pathologies' treatment. Nevertheless, due to different conditions such as: discomfort, that causes orthoses's removal, use them for less time than needed, use of footwear with the aim of orthosis replacement; the success can be impaired (29).

Nicolopoulos and his colleagues (29) stated the importance of scientific tests in order to evaluate the success of the orthotic treatment plan and assess the success or insuccess of their use, specifying the importance of biomechanical studies. The follow-up visits are very important in order to check the success or to provide any adjustments needed (30).

It is also important to perform investigation with unhealthy subjects since usually they are performed with healthy subject, which can conduce to wrong conclusions (29).

2.6.1. Ankle Foot Orthoses

Usually those orthoses cover the back of shank and the whole foot (26).

Rigid ankle foot orthoses are suggested in situations such as: weakness or absence of ankle dorsiflexors or plantarflexors, severe spasticity resulting from foot's equinovarus during swing or stance phase, weak knee extensors and proprioceptive sensory loss (26).

Hinged ankle foot orthoses allow free movement of the ankle in plantar flexion and dorsiflexion providing block of movement in coronal and transverse plane. Although presenting movement in the sagittal plane, these movements present some restrictions according to the range of motion available, which controls dorsiflexion and plantar flexion movement according to the patient needs (26).

Also preventing excessive dorsiflexion, those orthoses prevent tibia from collapsing over the foot, although allowing its movement forward over the foot (26).

Those kind of orthoses are helpful if the patient has a degree of eccentric control during second rocker, but it is necessary to prevent too much movement (26).

If the patient presents difficulties in plantar flexion, control and prevention of foot drop is needed during swing phase or if patient presents foot slap at heel strike, those kind of orthoses can also be helpful (26).

2.6.2. Insole

Some authors had already described effects of the insoles use on children with different disorders.

The use of insole (34) is helpful in the decrease of eversion and at the same time prevents further lower extremities injuries (28). Shih and Chen (34) justify it by referring that the use of insole helps the foot maintain neutral position, and it seems to have positive effects on injury treatment (28) (30).

Although, it is hard to find consensus over the authors. Some reasons are noticed by Stacoof et al. (2000) (33) as the result of their studies using different shape and material properties, testing also different kind of orthoses and soles, as well as presenting differences in methodological process as well as marker placements.

Shoe inserts cannot be successful by themselves (30), being important to select the correct footwear.

For those who tried biplanar insoles, there was a reduction on eversion, when comparing to the walking without insoles condition (28).

Various insoles' designs have been presented, including *the polysectional triaxial posting* method (29), a design that divides the foot in three areas: hind-foot, mid-foot and forefoot having separate posts in the different areas, which allows a better accommodation of foot deformities.

CHAPTER 2 – METHODS AND MATERIALS

1. Methodology

1.1. Study Type

This is a case study where a subject gait is analysed without any intervention done by the group of investigators, who only collected the data and produced reports about the data collected.

However, as further presented, different conditions were analysed and some components or conditions had been modified due to the results of each data collection, like what happens to orthoses between first and second data collections.

The subject was submitted to this gait analysis by suggestion of doctors from Hospital Dona Estefânia, where the child has regular medical monitoring. Doctors asked for our support with the biomechanical evaluation, since the Hospital does not have this kind of support system or specialists in this area.

This case is particularly interesting because of the lack of information about this disease, which turns to be relevant.

The subject's parents signed the informed consent once he is a minor (Appendix A).

1.2. Subject History

Without any suspicion of abnormalities, pregnancy was normal until the 34th week, when an *intrauterine growth retardation* was verified because the baby stopped gaining weight and because the presence of *oligohydramnios* was detected (that means the amniotic liquid was diminishing, which can be sign of other diseases (35)).

With the aim of letting the baby be born in due term, parents and medical group decided to wait two more weeks until the birth, checking the situation with echography supervision, almost daily.

The baby was born with 36 weeks, on February 20th 2008, 6:27 PM. He was born with 1750 grams and a cephalic perimeter of 31,5 cm, the reports accessed didn't mention the height at birth.

The weight and height were on percentile 5, the lowest one, and head circumference on the percentile 75, which is above normal.

Because of the lack of amniotic liquid and a caesarean surgery, he was considered a baby born in fetal distress.

After birth, the baby presented a good adaptation to life being considered a normal baby, however he needed to be in an incubator since he presented *icterus* and low levels of *glycaemia*.

Only at ten to eleven months of life some doubts about his development started to appear. Those doubts were based on his psychomotor development, and in the inadequate head growth, when compared to his body weight and height.

His doctor asked for a *transfontanellar ultrasound*, which reveals lesions all over the brain, more over the right side. After those results, it was necessary to do a *skull brain magnetic resonance imaging* and a *computerized axial tomography*. Those complementary exams revealed many lesions *supra and infra-tentorial*, including calcifications, being excluded an infectious cause.

Those lesions and the intrauterine growth retardation were significant to justify his diagnosis, which was done by the neurologist, Doctor José Pedro Vieira.

It was then necessary to evaluate his functions, make blood analysis, as well as an X-Ray.

The ophthalmologic study revealed signs of *severe vascular retinopathy* and calcifications.

The diagnosis, made around the ten months, was *Leukoencephalopathy, Calcifications and Quists*, a rare and complex genetic disease (there were no other known cases in Portugal at that time as well as nowadays), there was also a severe commitment of central nervous system and retina, with severe repercussions in terms of structure and functionality.

Summarizing, the first signs of this pathology were intrauterine growth retardation, unexpected proportion differences between body and head percentile, macrocephaly, marked motor delay, as well as huge hypertonia, and ophthalmologic problems. He has lack of motor coordination, which results of lack of muscular strength and lack of balance.

According to Professor Luís Nunes, the gene that causes this pathology was identified like gene *CTC1*, and on this specific case two mutations

of this disease-causing gene were identified. That were inherited from his parents.

At the same time that the etiologic investigation occurred, the baby started with ophthalmologic treatments to treat the lesions, and with early intervention with the aim of developing his psychomotor abilities.

During all those years, he had made regular *magnetic resonance imaging* and *computerized axial tomography*, as well as regular medical appointments and other treatments, especially ophthalmologic treatments.

Due to the rarity and specific characteristics of this disease, the parents searched for medical support outside Portugal. This search became truly necessary immediately after the diagnosis, when the ophthalmologist informed the parents that their child's situation was severe, because he was losing vision and he (the doctor) was not able to support it, as well as other doctors that were contacted in Portugal.

The first answers arrived from Badajoz and Barcelona. The baby had his first medical surgery in Barcelona with a known specialist, at international level, in this kind of specific problems.

After the ophthalmologic treatments and surgeries in Barcelona, different specialists in different clinics saw him, such as: Professor Eduardo G. Fernandes (Hospital da CUF Infante Santo) and Doctor Luísa Santos (Instituto Oftalmológico Dr. Gama Pinto). However, the best answer to the child's ophthalmologic problems was achieved at Intecir (Coimbra) with Doctor António Travassos.

The child had around 25% of his vision in left eye preserved, and his right eye is totally blind. That occurs in consequence of his neurologic

disease and it was one of the first consequences and disease characteristics.

The eyes diagnosis was confirmed on October 2009.

The clinic evolution was not positive, especially on at ophthalmologic level, with complete *amaurosis* of right eye, and the disease very widespread on the left eye, not stabilized, and needing permanent care.

Because of intestinal problems that can be associated with this disease and can be cause of death, helped by Professor Luís Nunes, on September 2010, the parents met Professor Amil Dias (Hospital São João do Porto). Professor Amil Dias accepted the challenge of introducing a capsule to film the *digestive and intestinal route*. The aim was to *screen for intestinal involvement caused by microangiopathy, the potential cause of gastrointestinal bleeding, anemia and osteopenia* – the results were normal without any signs of *microangiopathy*.

This child's motor development is also important to follow. He went from lying down to seated position, seating alone for the first time closer to 18 months old, starting to crawl between 18 and 19 months old, and his first steps occurred at the beginning of August 2009, achieving the independent walking in the end of October 2009 at 20 ½ months old.

When he was 15 months old, he was referred by his kindergarten teacher to the local early intervention team to have motor stimulation.

At 16 months old, he began intervention with a special education teacher and a physiotherapist, at that time three times in a week. Nowadays he still has physiotherapy, and according to his physiotherapist report now the physiotherapy occurs daily. The aim of his physiotherapy is to minimize the loss of functionality.

According to his family, nowadays he has also music therapy and hippotherapy.

The child presents clinic and neurologic evolution, however axial and trunk hypertonia as well as lack balance control and psychomotor development is on the inferior limit for his age.

His physiotherapist reported in the end of 2011-2012 (school year) that there is an increment of axial and limbs muscular tonus, with predominance on the left hemisphere, also presenting intention tremor of large amplitude which is reflected on his difficulty to do the activities that involve precision and the use of both hands at the same time. And he is able to realize independently the postural changes expected for his age.

About gait, the physiotherapist reported that his gait is asymmetric because of high tonus, with wide basis, foot pronation, knees in hyperextension, instable pelvis and lack of dissociation between waists (Fig. 1 and Fig. 2). That increases with speed increment, which can induce him to fall, being reported by his parents that at home sometimes he prefers to crawl instead of walking to avoid falls. Postural reaction to the lack of balance and arm protecting extension are present.

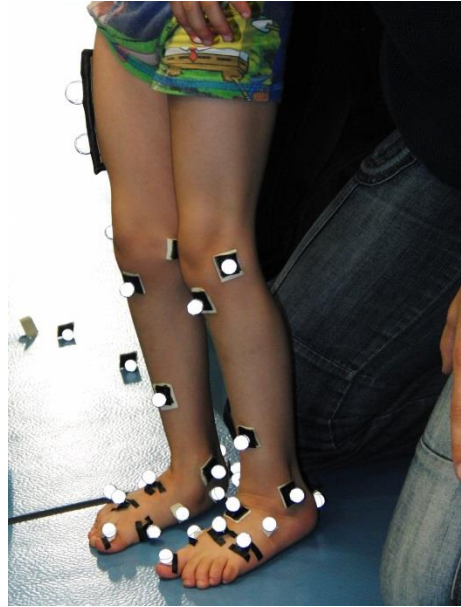
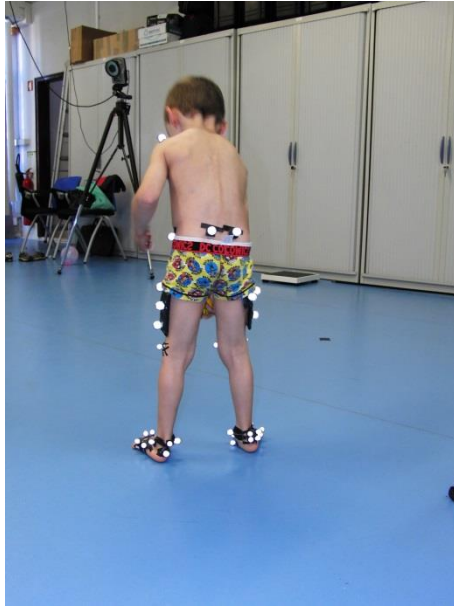


Figure 1 – Child photo static pose, posterior view (left) view between sagittal and frontal plane (right)



Figure 2 – Child photo static pose, lateral view (left) and feet (right)

Consequently, the child started to use dynamic orthoses in the tibiotarsal joints, to promote more distal stability.

Arising from parents' search for international help and support, the child is involved in two genetic investigation studies, one in Helsinki (Doctor Tarja Linnankivi) and another one in Manchester (Prof. Yanick Crow).

Beside those two doctors and their teams, others also accompany the child. For example, Doctor Jaume Campistol (neurologist at Instituto Dexeus), Doctor Marjo Van der Knaap (neurologist at VU University Medical Center), Doctor Rafael Navarro (specialized on genetic ophthalmology at Instituto de Microcirurgia Ocular de Barcelona), and Doctor Borja Corcostegui (specialized on ophthalmology more specifically in retina from Instituto de Microcirurgia Ocular de Barcelona).

Nowadays, the parents still have contact and share information and techniques with the Spanish specialists, Doctor Borja Corcóstegui specialized on retina and the genetic ophthalmologist Doctor Rafael Navarro.

In a recent report, the child's diagnostic was 91,24% of physical handicap. He is now 6 years old, but all the data collections were made between 4 and 5 years old.

3.3. Instruments for data collection and procedures

3.3.1. Information collected from medical reports and by interview

The subject's characterization, presented before, was made by reviewing the medical reports and complementary exams, and interviewing his parents (Appendix B) about the facts occurred during the child's life.

Firstly, the investigators received some reports, which briefly describe the child, as well as some of his characteristics, clarifying the pathological situation. Those were used to start his characterization, but it was necessary to involve the family in order to obtain more information that is not clear in the reports.

The investigators had been in contact with some of the doctors who follow the child regularly, or that have been in contact with him during those years, one of them is Professor Salomé Almeida, from the genetic department of Hospital Dona Estefânia. His physiotherapist had produced some reports and was present during data collections.

3.3.2. Instruments for Kinematic and Anthropometric data collection

Biomechanics is about movement, and according to Milner (36) the keystone is to measure and record the three-dimensional human movement. This kind of measurement and record needs a trained investigator who knows not only about the technical skills to deal with the system and ensures a good quality data, but also needs to know how to interpret the data collected.

Also according to Milner (36) according to the study that we intend to do, it is necessary to choose an appropriate marker system, which will be presented further ahead in this chapter.

The gait data for biomechanical analysis was collected at the Biomechanics and Functional Morphology Laboratory at Faculty of Human Kinetics, University of Lisbon.

Firstly we intended to collect kinematic and kinetic data, however some difficulties were found once we also wanted to preserve the child free walking speed as well as natural characteristics of gait. For those reasons, we were not in conditions to make the child step correctly over the kinetic boards. Because of that, we chose to focus our interest in the kinematic and spatio-temporal data.

We have taken into consideration that the hardware used to the kinematic data collection must be in agreement with the type of movement that we want to analyse, the more complex the movement the major number of cameras required (36).

In the present study, a system with 14 infrared cameras Qualisys Oqus 300, available in the laboratory, with a sampling frequency of 200Hz previously chosen in order to have a good resolution, was used. Those

cameras are in a high metal structure but if necessary can be moved, which happen during data collection, 4 were fixed to the tripods on the floor (Fig.3).



Figure 3 – Laboratory View

The Qualisys Oquos have a sensor resolution (pixels) of 1280x1024, 1.3 MP, a marker position resolution (subpixels) of 82000x65000, a maximum of 500 fps at full resolution and field-of-view, a maximum of 10000 fps at full resolution and reduced field-of-view, high speed mode at full field-of-view, 640x512 @ 1764 fps, active filtering for outdoor measurements, and high speed video support (37).

Those cameras are displaced all over the laboratory, equally spaced around a cuboid capture space, and for every data collection they were adjusted in order to reduce the dead space, like suggested by Milner (36).

Those infrared cameras are able to detect the reflective spheres placed (Fig. 4) under the set-up of anatomical points on the subject.



Figure 4 – Reflective Spheres used

In this study, a subject gait was studied, which involves the lower limb study, from which the setup of marks will be described after.

The capture volume was chosen according to the movement that we intend to analyse. In that sense, a hall (8 meters of length including turning area x 1.5 meters of width) (Fig. 5) was used, which passes over the small Kistler Force Platforms, placed on the laboratory to collect kinetic data, because in our first study we tried to collect kinetic data, and force platforms were synchronized with cameras.



Figure 5 – Hall used for Data Collection

The most basic body information is the length of segments between each joint (4). On every data collection with this subject, information such as age and the following anatomical information were collected: height (103cm), trochanteric height (49cm) and body mass (14,25Kg). This information was used to normalize gait data analyses on Visual 3D. In this specific case, and because of the child age as well as the

disability, it was our option to collect what was key information for our study, that is why we only collected this information.

The software used to analyse the collected data was Qualisys Track Manager for Windows XP® and Visual 3D for processing data.

3.3.3. Data Collection

In the total there were three data collections.

In the first one, the child enters the laboratory for the first time, and reveals to be a little shy and dependent on his parents' comfort, specially his mother's comfort. It was necessary to give him time to adapt to this new space. Also, to insure that he follows the correct trajectory without falling, which occurs frequently at this time, sometimes he walked with a hand support of one of the team members to give him more comfort.

He chose the velocity without any control by the team, and the hall is enough length to ensure that at least 2 meters of it were seen by the cameras.

The procedures: anthropometric measurement, markers placing, barefoot data collection, orthoses data collection, shoe data collection.

The team tried to register a good number of trials (considering that a good trial is a trial in which the child/markers are clearly seen by every camera) which are enough to compare the different conditions in study: barefoot, orthoses with shoe, shoe and insole with shoe (this one collected only in the last data collection).

In the second data collection, the child looks more comfortable in the laboratory and with the team, cooperating more. This time it was proposed to him to play a game (Appendix C) and this game used the same hall used the first time.

The procedures were the same used in the first data collection: anthropometric measurement, markers placing, barefoot data collection, orthoses data collection.

In the third data collection, the child looks a little uncomfortable because now he knows the pain associated with taking markers after the data collection, so he rejected the markers placing, finding ways to take them fast. With his parents' cooperation, it was a little easier to keep the markers. The same hall was used, the same game, and the procedures were the same: anthropometric measurements, markers placing, barefoot data collection, insoles data collection, and orthoses data collection.

The team have always collected a static trial, that was dependent of the child's availability (mood) for this kind of performance, always trying an interval of time of 30 seconds to one minute collecting the maximum time of static position to ensure 10 to 15 frames to build the model in the Visual 3D.

After the dynamic collection, that for each condition took approximately 2 minutes, and dependent of the child's execution, 2 or 3 trials were collected.

3.3.3. Anatomical Points

Markers attached to the body are usually placed to represent our best estimate of a joint centre (4) and over a body anatomical landmark close to the centre of rotation of a joint (26).

In the present study, passive markers were used to indicate the position and orientation of the body in three-dimensional space (36). Those markers are visible by infrared cameras because one of their properties is being reflective spherical markers. Those reflections are detected by the cameras in the laboratory, recording the body movement.

Clinical gait analyses require a model which can be applied to patients according to their age or cognition and which uses instrumentation that is not affected by gait pathology (5).

The set up was created according to some criteria described as follows.

Markers with 1 cm or 2 cm of diameter, depending on the surface and number of markers needed in the segment, for example for feet small markers were used and the larger ones at clusters.

Markers were placed on lateral and medial aspects of joints on anatomical landmarks at proximal and distal ends of the segment, and additional clusters were placed on each segment, thus the anatomical landmark markers enable the proximal and distal ends of the segment to be identified in relation to the clusters markers (26).

Clusters are three non-collinear markers placed on the thigh and shank to define their position and orientation in three-dimensional space (36).

The use of clusters is necessary to provide static trial to locate the anatomical points relative to the cluster.



Figure 6 – Set up for barefoot condition



Figure 7 – Set up for orthoses with shoe condition

Clusters should be at least three non collinear markers in order to track not only the segment position but also its orientation, in six degree of freedom, which means in sagittal, coronal (frontal) and transverse planes (26).

The calibrated anatomical system technique also known as CAST, proposed firstly by Cappozzo et al (1995), has a huge contribute towards standardizing movement description in research labs and clinical centres for the segments pelvis and lower-limb (26).

It does not matter where cluster is placed since CAST technique uses the relative positions, being useful as tracking markers. In order to be

used as tracking markers, it is suggested that those clusters should be placed at an angle between coronal and sagittal planes (26), during the present data collection clusters were used for thigh, leg and foot.

The setup of markers was chosen according to literature review, Oxford Foot Model, adapted to the child specificity and conditions (Fig. 6 and Fig. 7) (26) (38): right and left anterior superior iliac spine, right and left posterior superior iliac spine, right and left lateral knee, right and left medial knee, right and left lateral malleolus, right and left medial malleolus, right and left posterior calcaneus, right and left calcaneus tuberosity, right and left 1st and 5th metatarsal head (distally), right and left 1st and 5th metatarsal head (proximally), right and left hallux (Appendix D).

The traditional single-segment, also known as rigid model used for the foot in clinical gait analysis and human movement research, is starting to be replaced by different models in which the foot is divided into multiple rigid segments (38). The use of 3D multisegmented foot models is popular in gait laboratories as it would seem to be an adequate tool for the *in vivo* analysis of dynamic foot kinematics (39).

Foot markers are not defined so simply as the rest of the segments once it was constructed as a multiple segment, since pathological conditions could be better studied with a model that divides the foot in the three different parts that compose it (26).

However, when talking about the orthoses and shod foot, even using the same model, in those two conditions, markers aren't attached to the foot which means that a lot of artefacts can be considered, and we cannot be sure of the foot position when shoe (26). The multisegmented foot is easier to use for barefoot condition than for the other conditions (26).

Some markers were used specifically for the calibration (medial knee and medial malleolus). Those markers are necessary to the orientation of the axys system of the three tracking markers, helping define its orientation. During data collections no one took that out, because it did not have bad influence on gait, and in order to reduce boy's anxiety about taking the markers, which is painful for him.

In order to minimize the movement artefact and to be fast and easy to attach markers to the child's body, they were attached directly to the skin, but also to the orthoses and to the shoe, according to each condition evaluated, with double-side tape, reinforced with more tape over.

It's recommended that only a person places markers in order to remove inter-individual variability as a source of error (36) so a person from the team was chosen to do it.

3.3.4. Calibration

Calibration should be done before each data collection session, even if cameras were not moved after the last session (36) but every time that something that can modify calibration happens (example: if someone kicks a camera) they must be calibrated again.

The kinematic system calibration was made in agreement with Milner (36), in two phases, first static calibration (or seed) and second dynamic calibration (or wand).

The system was calibrated in order to be free as much as possible from noise and artefacts (4).

The space used was a hall with 8 meters of length including turning area x 1.5 meters of width.

For the static calibration, a rigid L-frame was used, with four markers in known locations. The L-frame defines the location of the origin and the orientation of the laboratory reference frame and the force platforms. This is a way to have standard definitions and helps in the communication between laboratories (36).

It is important to establish a convention system in order to keep track of all the kinematic variables (4). The use of a system convention will allow describing a movement in a common language. To describe the segment in reference to ground or gravity direction, an absolute spatial reference system named Global Reference System should be established, saying that GRS presents the coordinates and is fixed in the laboratory or data collection space (4).

It is also possible to have a Local Reference System and Rotation Axes (4) within each segment, the anatomical axis system is set with its origin at the center of mass of the segment.

4 markers of 1 mm each were also used, since were the smallest used in data collection, and were displaced as close as possible to ensure that small markers in a closer space, such as his foot, were seen by cameras, like suggested by Richards (26).

After calibrating the whole space, every camera was checked for the need of being adjusted in its sensitivity, to ensure that only the markers were seen and each mark with the highest quality possible.

After the static calibration, a dynamic calibration takes place in order to ensure that all the volume needed to data collection is calibrated. This is the direct measurement of an object with known dimensions made by all the cameras throughout the entire volume capture (36).

The force platforms for kinetic data collection were used during the first and second data collection and those platforms did not need calibration since they were already calibrated.

According to the International Society of Biomechanics (Wu & Cavanagh, 1995 (36)) the X axis must be the direction of progression, the Y axis the vertical, and Z axis the mediolateral.

If considering the Cardan sequence: x-axis is in the medial-lateral direction, y-axis is anterior posterior (direction of travel) and the z-axis is the up and down or axial direction. So if talking about joint coordinate system: x is about flexion/extension, y is about abduction/adduction, and z is longitudinal internal/external rotation (26).

In the present study the following was considered: X represents direction of progression, Y represents mediolateral, and Z represents vertical.

Angles have a zero reference and a positive direction (counter clockwise means positive angles) (4), the same for velocity which is positive when increasing.

3.3.5. Data Processing

Firstly representative trials with a complete gait cycle were selected, and for each one markers were identified correctly, in each frame, joining broken trajectories and deleting unnamed markers or ghosts, like suggested by Milner (36).

Joining broken trajectories in Qualisys Track Manager can be an option. Because data were exported to Visual 3D, in c3d format, it wasn't necessary to do gap-fill, because Visual 3D is able to complete trajectories, so if Qualisys Track Manager automatically adds gap-fill,

trajectory was checked, to verify if it was correct, for other situations gap-fill wasn't used.

Visual 3D was used to data analysis. Visual 3D allows the user to import motion capture and force data collected. In the present study, data were collected using Qualisys Motion Capture System and exported in c3d format (40).

Once with data added to Visual 3D workspace, it is possible to perform data computations, filter and transform data building into biomechanical models, performing analyses, and building reports (40).

Visual 3D_Pipeline is a set of commands that are processed in sequence, used to automate processing steps. The pipeline has the ability to manage files, define events, execute signal-processing computations, create and edit models, create and modify reports and generate statistics (40).

In Visual 3D there is no standard coordinate system, like the one used in the motion analysis community, because authors assume that each individual user has a standard coordinate system for their laboratory (40).

As default, in the Visual 3D Coordinate System (V3CS), the global axes are oriented such that V3CS Z axis points vertically upward, the V3CS Y axis points along the direction of progression and the V3CS X axis is perpendicular to the other two axes as described by a right handed Cartesian coordinate system (40).

A Virtual Lab was created because there was the need to *represent the floor so that motion capture markers can be projected onto the floor* (40), being a way to represent the Laboratory with a well-defined Segment Coordinate System.

As recommended by Visual 3D (40), to start any of the three data analysis, a C3D file of each static pose for condition in the three data collect was added to it.

After, a chosen number of C3D files for movement trials, in equal number for each data collect was added. In the first data collection three were added, in the second data collection eight were added and finally, on the third data collection twelve were added. This difference results from the child's performance.

Then the team created a model template, and the signal-processing pipeline, with the events heel strike and toe-off for each foot, and a report template.

Data was normalized by subject mass.

The model was created according to the markers placed under the anatomical points and defined according to the proximal and distal markers defined for each segment and the tracking marks for each segment. In case of pelvis, CODA Pelvis model was used.

After creating the model, movement files were open/added, indexing them according to the condition in evaluation (40) and missing data points were interpolated in order to create the signals.

A filter was added taking into account that there are different kinds of filters resulting from different techniques and from which results differ somehow. The most common technique used to attenuate the noise, which is signal appearing in the data that becomes from artefacts and not from the real signal, is digital filtering, a low pass filter. It is a noise attenuation technique based on differences in the frequency content of the signal versus the noise (4).

According to Milner (36), noise due to skin marker movement is generally of high frequency, and the human movement occurs usually at low frequency. If using a low pass filter, it is possible to separate the components of time displacement curve of a marker based on whether the components oscillate above or below a chosen cut-off frequency, and discard all components above the cut-off. It is more efficient to use a cut-off frequency according to the different movements, which represents a filter at 6 Hz for walking, so a Low Pass Filter of 6 Hz was used just like suggested in literature for this kind of data.

After, we have created events: if possible (if the child passes over the platform correctly) we used platform signals to determine events, if not we did it manually: Heel Strike and Toe-Off, from right and left, and every time that we have force platform signal we also select On and Off of the platform.

All variables were time-normalized to percent gait cycle.

3.4. Variables Operational Definition

Dependent variables: spatio-temporal parameters and kinematic parameters

Determinant independent variables: orthoses with shoe and insole with shoe

Confounding independent variables: subject's age, gender, body mass index

CHAPTER 3 – RESULTS & DISCUSSION

1. Results Presentation and Discussion

The present results, collected at three different times, have as their main aim to observe and compare the child's gait patterns in different conditions, and different periods to describe gait development.

In the first data collection, that took place on 11th January 2013, the following conditions were compared: barefoot (Fig.8), orthoses (DAFO Tami PF Block, Fig. 9) with shoes, and a pair of shoes (Fig. 10) that the child usually wore.



Figure 8 – Barefoot condition

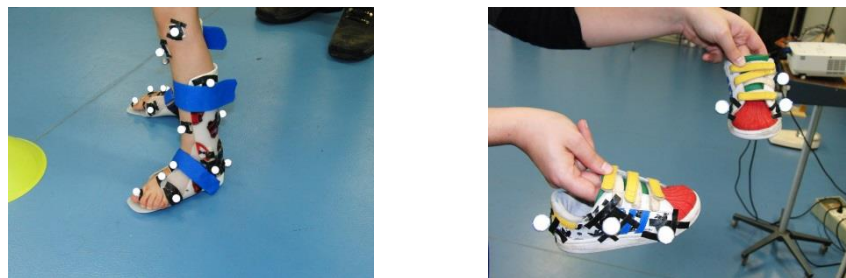


Figure 9 – Orthoses image before getting into the shoe (left) and shoe (right)



Figure 10 – Shoe condition

The second data collection took place on 22nd March 2013, almost one month after the beginning of modified orthoses's use. Those modifications were made in order to add range of motion to plantar flexion and dorsiflexion, when using it. Information was collected in the following conditions: barefoot (Fig. 8) and orthoses with shoes (the same used in the first data collection but adjusted, Fig. 9).

In the third data collection, that took place on 19th July 2013, almost one month after the beginning of insole's use, information on the following conditions was collected: barefoot (Fig. 8), orthoses (the same used on second data collection, Fig. 9) with shoe, and insole with shoe (Fig.11).



Figure 11 – Insoles

Specialists that do not belong to Biomechanics and Functional Morphology Laboratory's team have recommended orthoses and insole. The child's parents consulted those specialists themselves.

During the data collection, the team from the Biomechanics and Functional Morphology Laboratory observed that this child doesn't present a regular walking pattern, when compared to literature review, since sometimes he uses heel in the initial contact (with the floor), but other times he does a toe contact.

The toe contact was observed during the first data collection although there were few cycles and sometimes due to the falling characteristics of his gait.

During the last data collection, that was observed again, especially when walking with insole with shoe, which can represent a toe walking pattern (41), although this can be an occasional situation due to the adaptation to insole with shoe walking, which is described by the child as "walking unlike", meaning that it is an important point to observe in the future evaluations.

Following, the achieved results presented by data collection can be seen as well as the discussion of the same results.

1.1. 1st Data Collection: 11.01.2013

1.1.1. Temporal Distance Results ($\bar{X} \pm \delta$)

		Barefoot	Orthoses+Shoe	Shoe
Speed (m/s)		0.804	1.053	1.098
Stride Width (m)		0.088±0.010	0.088±0.026	0.078±0.039
Stride Length (m)		0.809±0.043	1.038±0.046	0.961±0.100
Cycle Time (s)		1.007±0.142	0.986±0.028	0.875±0.048
Step Length (m)	Left	0.377±0.0057	0.516±0.031	0.471±0.069
	Right	0.435±0.047	0.532±0.031	0.474±0.051
Step Time (s)	Left	0.490±0.086	0.483±0.009	0.427±0.019
	Right	0.516±0.066	0.493±0.028	0.454±0.038
Stance Time (s)	Left	0.607±0.063	0.560±0.044	0.502±0.050
	Right	0.600±0.129	0.530±0.041	0.440±0.016
Swing Time (s)	Left	0.382±0.071	0.419±0.016	0.388±0.021
	Right	0.426±0.039	0.448±0.037	0.429±0.031
Stance Time / Cycle Time (%)	Left	≈ 60.3	≈ 56.8	≈ 57.4
	Right	≈ 59.6	≈ 53.8	≈ 50.3
Swing Time / Cycle Time (%)	Left	≈ 37.9	≈ 42.5	≈ 44.3
	Right	≈ 42.3	≈ 45.4	≈ 49.0
Double Limb Support Time (s)		0.203±0.091	0.103±0.043	0.064±0.019

Table 2 – Temporal Distance Results 1st Data Collection

Regarding the **speed** values, it is possible to observe that, between conditions, speed increases, so the highest speed value achieved during the data collection was with shoe and the lowest on barefoot data collection.

Stride width values are lowest in shoe condition when compared with the other two: barefoot and orthoses with shoe, which are almost equal.

Stride length is higher for the condition orthoses with shoe than the other two conditions. Barefoot has the smallest stride length value.

About **cycle time**, the highest value was for the barefoot condition and the lowest value for the shoe condition.

Regarding the **step length**, it is possible to observe that the highest values obtained were when using orthoses with shoe. Barefoot

condition has the lowest values and the highest difference between left and right feet.

However, for all conditions, left stride length was smaller than right stride length. The same happens for **step time**.

Stance time is always higher than **swing time** (%); nevertheless not in the proportion reported on literature which is 60% (stance) to 40% (swing), which is in agreement with the child performance once that sometimes it seems that he was walking like falling repeatedly.

Double Limb Support presents higher values, meaning more time with feet on the floor, for barefoot condition compared to the other two conditions.

Despite all the information above, in a general way and according to the data collected comparing barefoot condition with the other two conditions, it is possible to observe that barefoot is the condition with worse results, followed by orthoses with shoe condition, since it has less speed, small stride length, higher cycle time, higher double limb support. This can mean that when walking on barefoot, the child does not feel so comfortable, and reporting to the visual impairment chapter, he takes more time to analyze the ground and organize himself to walk forward. It is also known that shoes in contact with the ground have friction which can help him to feel more comfortable/fearless, however shoes do not let him explore the ground as barefoot, meaning that he can be more focused on walking forward instead of getting information from the floor (19) (18).

1.1.2. Intersegmental angle on sagittal plane

1.1.2.1. Ankle

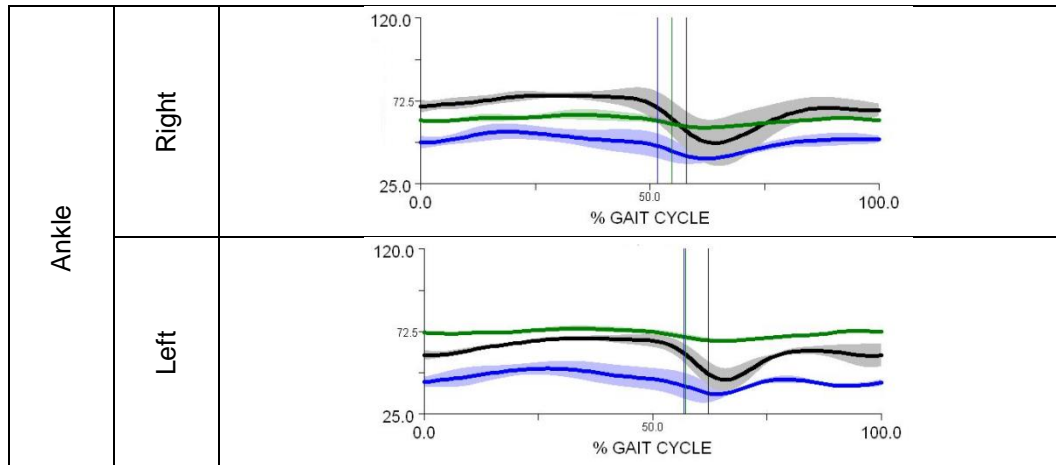


Table 3 - Ankle intersegmental angle on sagittal plane (dorsiflexion [+], plantar flexion [-]) Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.

In a regular gait, after the contact of foot with the floor, a plantar flexion is observed, which is not possible to observe on the graphs presented on the table above. That is not possible to observe since one of the child's difficulties is the movement between dorsiflexion and plantar flexion. Observing the images collected, during barefoot collection it is possible to observe that the child hits the floor with the entire foot, which is expected because of his movement difficulty, but it is also a characteristic of children with low vision, who tend to contact the floor with all the foot to collect more kinesthetic information (18). Orthoses with shoe condition and the shoe used during this collection add some restrictions to this movement execution.

During unilateral stance phase, for regular gait cycle, we are supposed to observe ankle moving in dorsiflexion until the maximum of 50% of gait cycle. Looking to the graphs, it is possible to observe that for all the conditions this does not happen, being compensated (as will be possible to observe next) by the knee flexion which is also described as an adaptation (25).

However, for barefoot condition comparing to the other two conditions, it is possible to observe dorsiflexion happening but only after toe-off. Orthoses with shoe condition do not present clear movement between dorsi and plantar flexion during the total cycle.

During this collection, it was not possible to observe significant range of motion between dorsiflexion and plantar flexion. Barefoot is the condition that presents a more evident movement; however, shoe condition presents it but it is a dorsiflexion's smooth increase that finishes earlier than barefoot dorsiflexion.

In the final phase of stance, plantar flexion occurs again, being more evident to barefoot condition. After, ankle moves again in dorsiflexion.

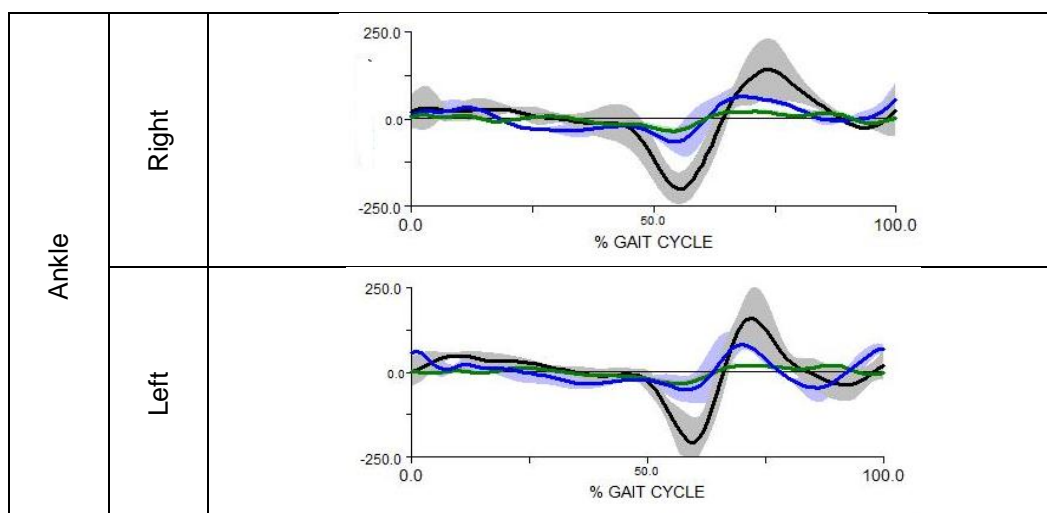


Table 4 – Ankle velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

It is expected to observe lower angular velocity on the ankle when compared to knee and hip.

Ankle presents maximum velocity right before the instant when the foot leaves the floor, approximately at 60% of gait cycle, as presented in both graphs.

The highest joint velocity is the one that corresponds to barefoot, which has the highest range of motion. Orthoses with shoe velocity is almost null also corresponding to the intersegmental angle presented.

Barefoot condition seems to be the closest to regular situations.

1.1.2.2. Knee

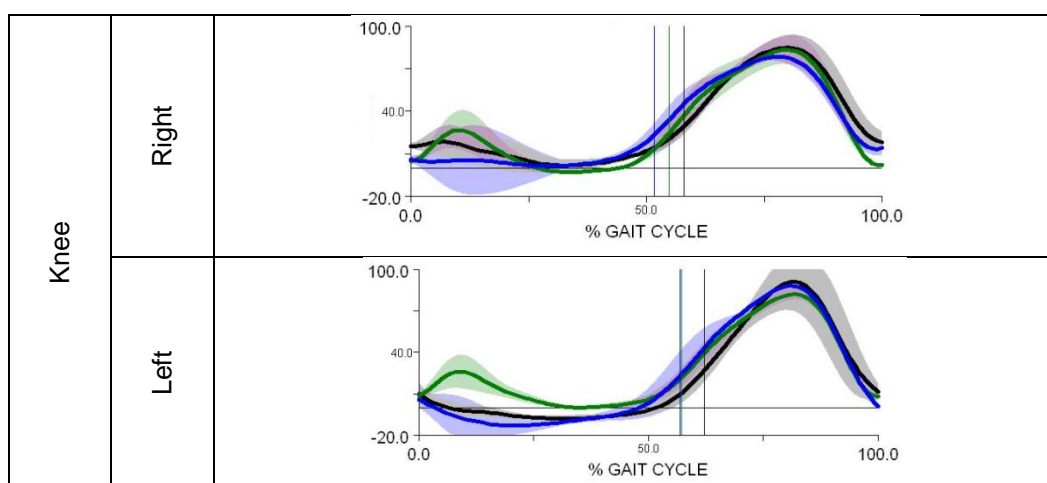


Table 5 - Knee intersegmental angle on sagittal plane (flexion [+], extension [-]).
 Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.

When heel strike occurs, in a regular gait cycle, knee should be on extension, which happens at approximately 10% of the cycle, moving to flexion until the end of double support.

Observing the graphs above, this is evident for orthoses with shoe condition, and it is possible to observe that this condition is the closest one to the expected for regular situations.

It is also possible to observe that shoe condition does not present the expected curve, and even that barefoot condition presents a small flexion's peak for right knee, left knee does not present it, being similar to shoe condition curve. Observing left knee, it only presents this first

flexion curve when orthoses with shoe condition take place, the other two do not present it.

At approximately 40% of gait cycle, an extension movement should occur, which is possible to observe for orthoses with shoe condition very clearly.

After knee returns to flexion, which happens closer to the instant when the foot leaves the floor, this movement presents a marked peak in a regular graph and here for all the conditions, even that occurring earlier in some cases than in others, it is also possible to observe it.

After this flexion peak, knee returns to extension, and finishes the gait cycle in a total extension, which is observed in all curves.

Orthoses with shoe condition present the movement pattern closer to the described on literature despite the ankle dorsi plantar flexion restrictions, orthoses with shoe condition present better performance on the knee movement than the others.

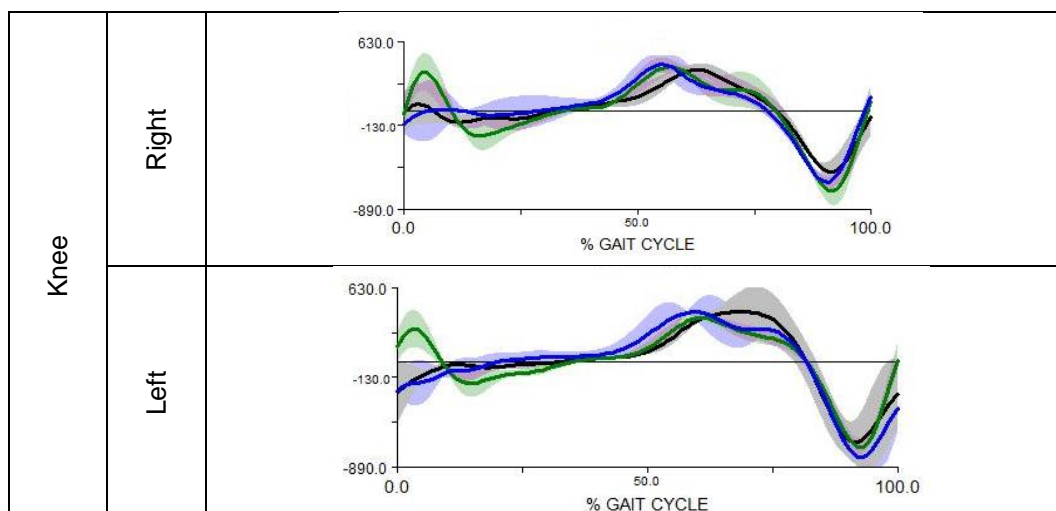


Table 6 - Knee velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

It is expected that knee presents higher angular velocity when compared with ankle and hip.

Knee presents maximum velocity when the foot leaves the floor, and on the final phase of swing, approximately at 90% of gait cycle, like presented in the graphs above.

Angles and velocities match.

1.1.2.3. Hip

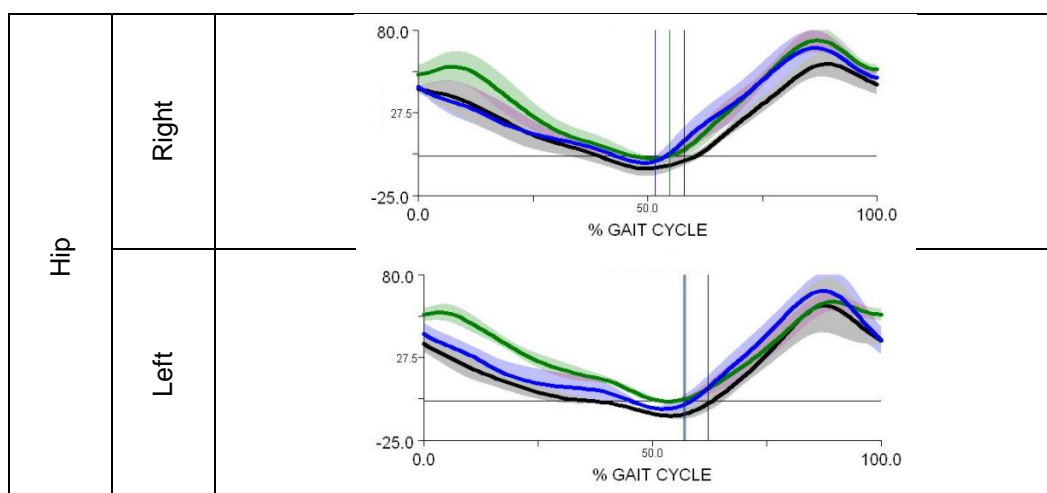


Table 7 - Hip intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – Ortheses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.

At initial contact, when foot starts the contact with the floor, hip is on flexion, like presented in the graphs above, for all conditions. After, when the final phase of stance is reached, an extension movement occurs.

Then hip returns to flexion, which is also obvious in the graphs for every condition, although it seems to start earlier for right hip than for left hip.

In a general way, hip movements are in agreement with the expected.

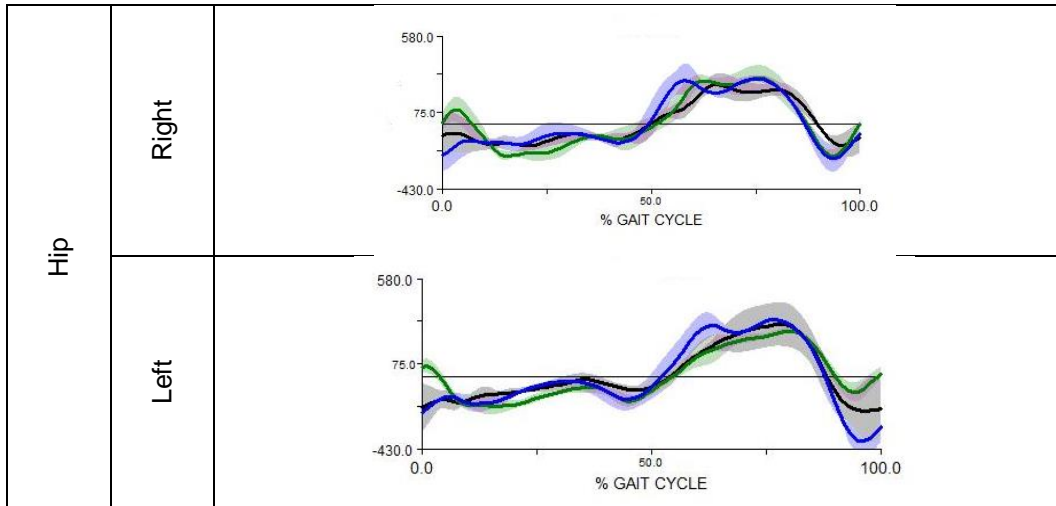


Table 8 - Hip velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

It is expected to observe hip's highest angular velocity during the initial phase of swing, approximately at 70% of gait cycle. At the same time, velocity at knee should be null, like presented in the graphs above.

1.1.2.4. Pelvis

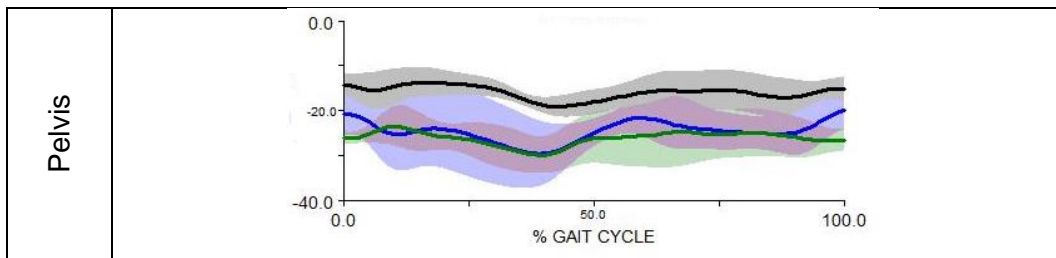


Table 9 - Pelvis intersegmental angle on sagittal plane (anterior flexion [+], posterior flexion [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

When analyzing pelvis on sagittal plane, pelvis tilt is being analyzed, meaning the movement between anterior and posterior flexion.

The pelvic movements, on this plane, consist in small variations in the degree of anterior pelvis tilt, which should be inclined downwards anteriorly (3). However, observing the graphs, there are small variations but pelvis is posteriorly, representing the child's movement, which can be the result of the fear of falling and a way to control his body.

It is desirable to obtain two peaks and two valleys. The valleys take place when toe-off and opposite toe-off happen, and peaks occur at late single stance and mid-to-late swing phase (3), although that is not what is possible to observe from the graphs obtained. Peaks and valleys are not obtained in the expected instants, and barefoot presents along the gait cycle small curves that are not expected.

1.1.3. Intersegmental angles on frontal plane

1.1.3.1. Ankle

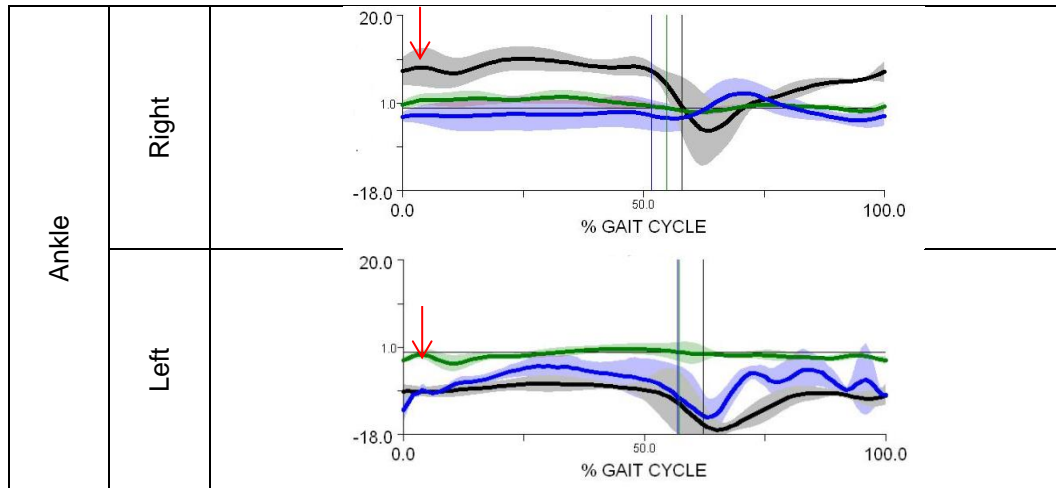


Table 10 - Ankle intersegmental angle on frontal plane (eversion [+], inversion [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.

The child's feet are usually in eversion (static pose) and the use of orthoses has as one of its aims to correct it.

During the initial contact of foot with floor, an eversion movement of the ankle should occur, but observing the graphs above, for all the conditions that is not clear, and can be justified by the feet's eversion observed under static pose. Nevertheless it is possible to observe the referred eversion peak in some data (red arrow), being more obviously for right barefoot and left orthoses with shoe conditions.

The use of orthoses will cause limitation on movement (26), especially on eversion and inversion. This is why, observing the graphs for orthoses with shoe condition, it is possible to observe an almost straight line describing the movement, meaning that ankle was like stable, without movement.

It is also possible to observe in the graphs, for barefoot and shoe conditions, two things: inversion occurring too late in time, since it was

expected before, and eversion occurring instead of inversion. It is possible to observe that right barefoot, left barefoot and left shoe present inversion movement like expected.

Then an eversion movement happens, helping the foot returning to neutral position, which is possible to observe for shoe and barefoot condition, this last one better for left foot than right foot.

Barefoot seems to be the closest to normal gait cycle although orthoses with shoe correspond to the expected for that condition (almost total absence of movement).

1.1.3.2. Knee

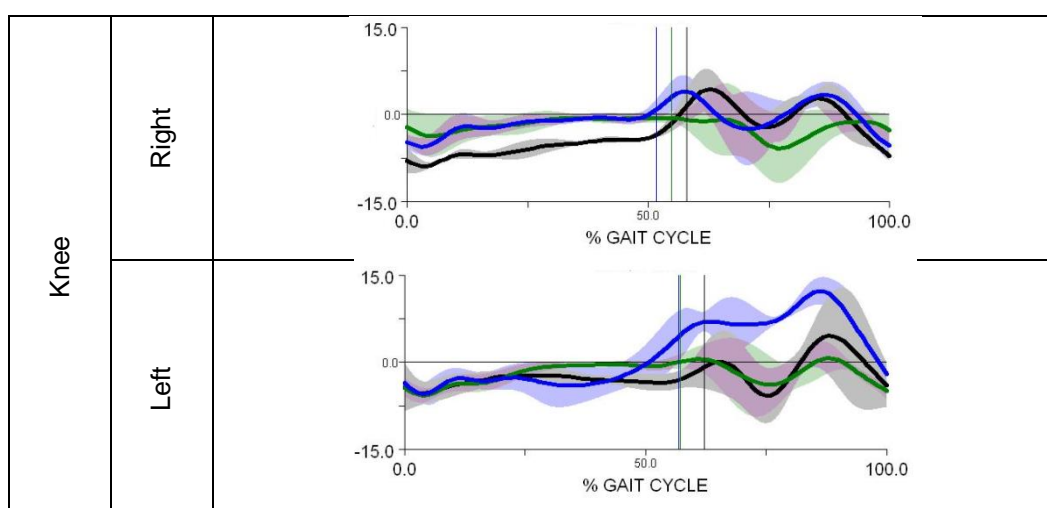


Table 11 - Knee intersegmental angle on frontal plane (adduction [+], abduction [-]).
 Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.

During the literature review about the child's disease, one of the characteristics referenced is the knee's *valgus* (1), which has influence not only on the knee but also under the proximal joints that work in chain with knee, just like stated before both feet when in barefoot static pose are in eversion.

From the images collected, even when some of them present the knee's *valgus*, that is not as obvious as everted foot.

Like suggested on the literature review for knee under frontal plane (4), it is expected to observe stability because of joint restrictions, although small movements occur between adduction and abduction.

During stance phase, knee should be in adduction, and the range of motion should increase until about 60% of gait cycle. After, during the swing phase, knee changes to abduction until the end of gait cycle.

Looking to the graphs above, it is possible to observe that orthoses condition does not present the expected movement that can be justified by the fact that the foot is being corrected by the use of orthoses and that can promote knee's abduction.

Barefoot condition also presents an irregular pattern, changing from abduction to adduction (after toe-off) and again to abduction, the same happening for right foot of shoe's condition. Left foot of shoe's condition, even if presenting an irregular curve when changing from abduction to adduction, is the closest to regular curve.

1.1.3.3. Hip

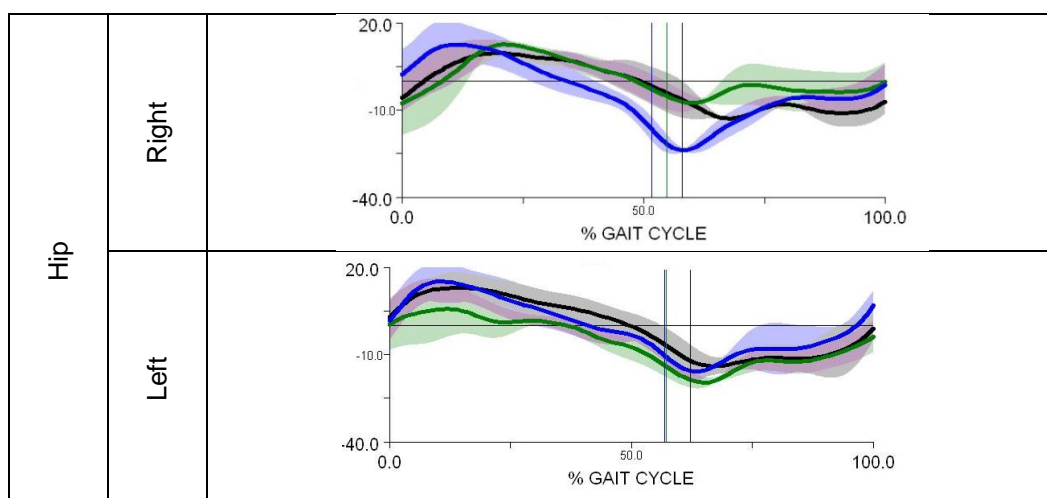


Table 12 - Hip intersegmental angle on frontal plane (abduction [+], adduction [-]).
 Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe. Vertical line represents the instant when foot leaves the contact with the floor.

During the initial contact, it is supposed to be observed hip with 0° passing to abduction with approximately 6° , at the end of double support phase. Observing the graphs, it is possible to see that a small increase on hip abduction occurs.

After, hip should move slowly into adduction, finishing the movement at about 60% of gait cycle. Adduction occurs for all conditions, although not very obvious for all, which can be justified with the child's immature gait and also the visual impairment, both have associated decreased hip adduction in stance (25).

Then it should return to abduction, which occurs in every condition.

First data collection presents a pattern of movement, closer to the regular curves described on literature.

1.1.3.4. Pelvis

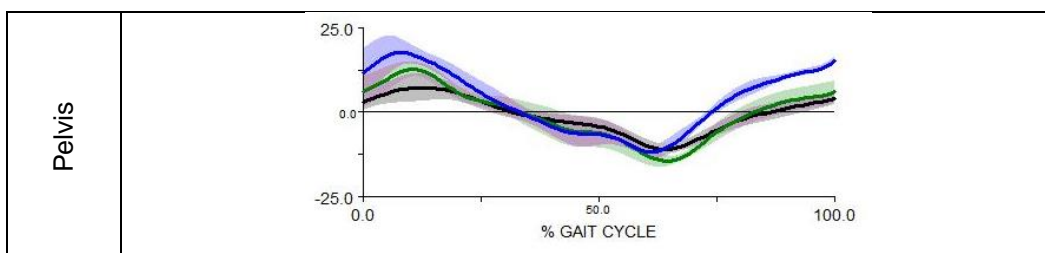


Table 13 – Pelvis intersegmental angle on frontal plane (Up [+], Down [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

In the frontal plane during early stance phase the contralateral side of pelvis drops downward. The peak of pelvic obliquity occurs right after opposite toe-off, which corresponds to early stance phase on the weight-bearing limb, like is possible to observe on the graph above presented (26).

After it drops, reaching a low point halfway through single stance, followed by a smaller peak elevation when occurs opposite foot-strike (3).

The peaks are correlated with opposite toe-off and opposite foot-strike.

1.1.4. Intersegmental angles in transverse plane

1.1.4.1. Ankle

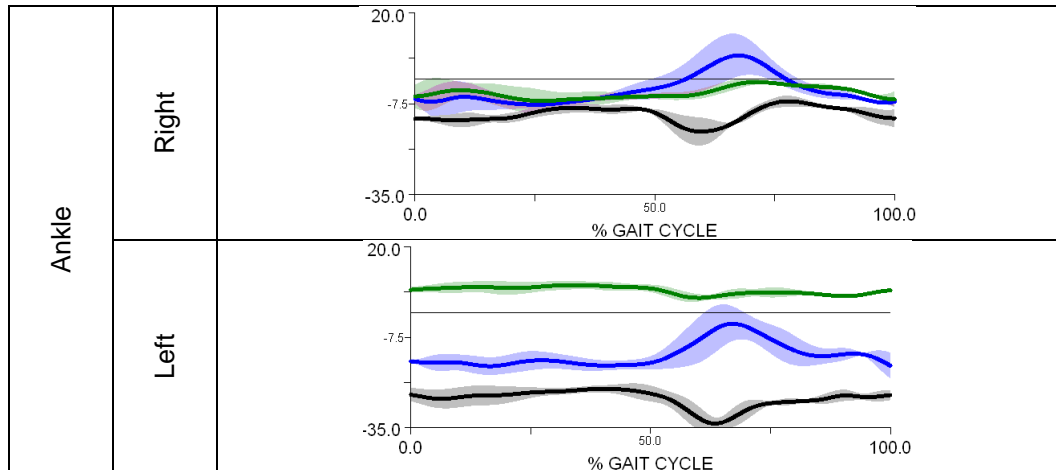


Table 14 - Ankle intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe.

In the beginning of initial contact, foot should be on neutral position or external rotation, followed by a decrease in the external rotation degrees during the period when foot establishes contact with the floor, which occurs during elevation of contralateral foot.

The use of orthoses will cause a limitation to those movements (26). That is why, observing graphs, it is possible to see that orthoses with shoe condition do not present relevant motion changes, it goes like a straight line along the gait cycle. However, it is expected to observe both feet in external rotation, and the left foot is always in internal rotation which can be influenced by eversion movement.

Barefoot condition presents noticed external rotation peaks after 50% of gait cycle. It is possible to observe that, when barefoot presents external rotation peaks, shoe presents the opposite, internal rotation peaks.

Barefoot condition seems to be the closest one to a regular gait cycle, even when the movement degrees are not the expected and the curve is the closest one.

1.1.4.2. Knee

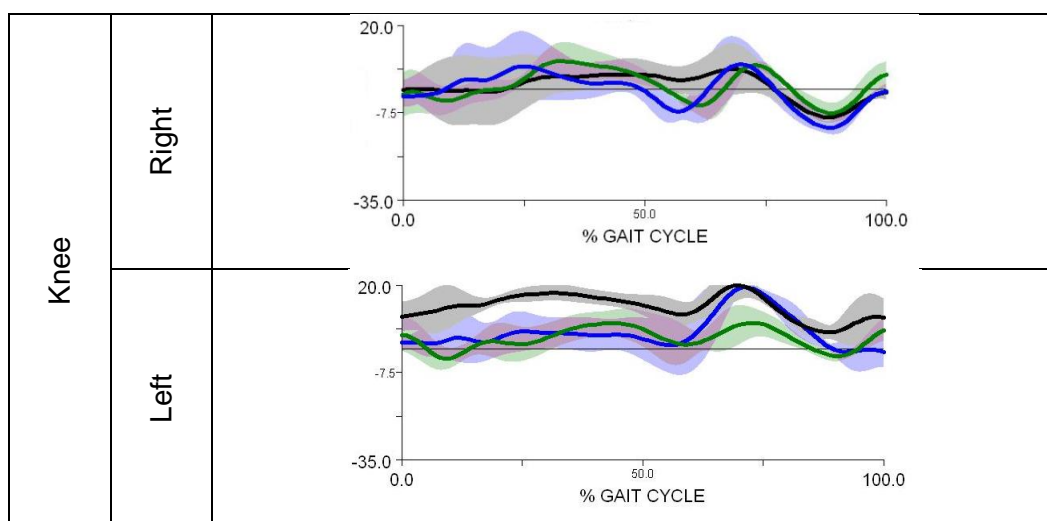


Table 15 - Knee intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot; Blue – Shoe.

Knee rotation is linked to flexion and extension movement, so usually knee is in external rotation during gait cycle, only modifying the range of motion along that, being possible to observe internal rotation after foot strike.

What graphs above show is that during right gait cycle, until 50%, knee is in internal rotation with two peaks of external rotation after that. It is also possible to verify that left knee only presents two smooth peaks of external rotation, being at internal rotation during the rest of the gait cycle.

This can be due to the *genua valgus* (1) reported for this child's disease, which contributes to the internal rotation, and also to the feet eversion.

1.1.4.3. Hip

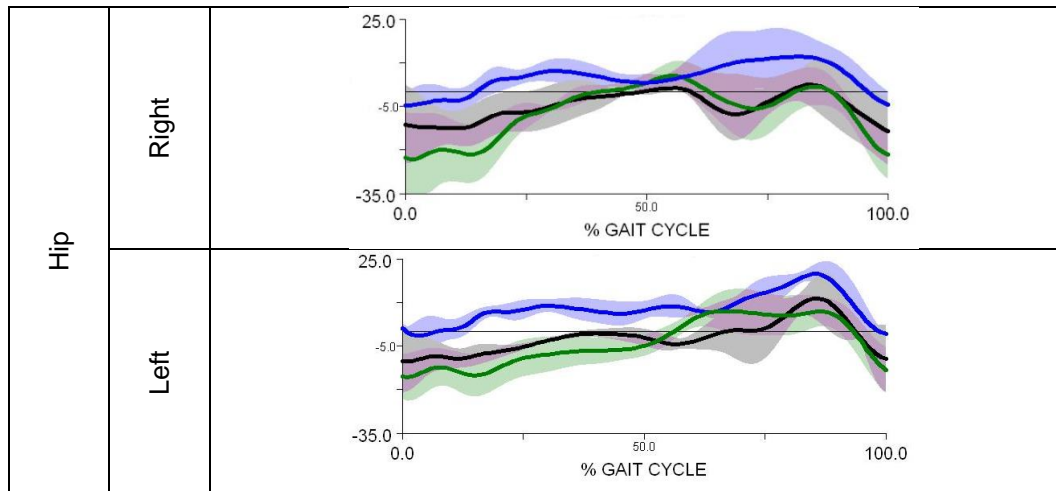


Table 16 - Hip intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

In normal situations, during initial contact, hip rotates externally, but rapidly starts to rotate internally, returning to external rotation at thereabout 70% of gait cycle and rapidly moves again into internal rotation.

For all the conditions evaluated, except for left shoe, when the foot strikes the floor, the hip is on external rotation.

Shoe condition presents internal rotation in the following instant. That is more difficult to observe for barefoot and orthoses with shoe conditions, and worse for right hip that only presents two smooth peaks after 50% of gait cycle for barefoot and orthoses with shoe condition.

Left hip presents internal rotation movement for barefoot and orthoses with shoe condition starting after 50% of the gait cycle and until about 85%, and all the conditions finish the gait cycle in external rotation.

1.1.4.4. Pelvis

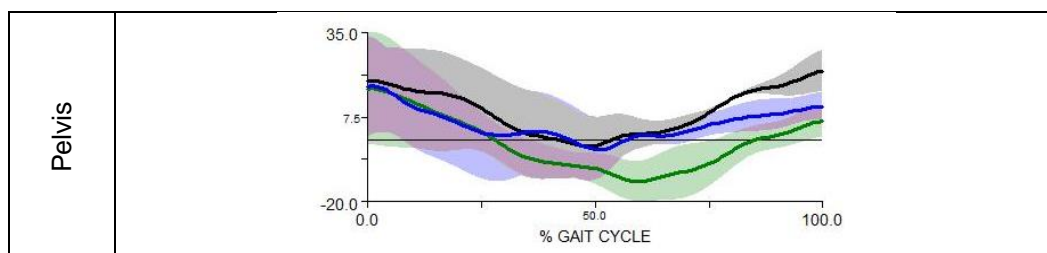


Table 17 - Pelvis intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot; Blue – Shoe.

The peak of internal rotation occurs slightly after foot-strike (3) and pelvic rotation decreases in a faster way when heel strike occurs and is reversed when full weight is placed upon the foot (2), as stated on the graphs above.

With load acceptance, the pelvis begins a counter-rotation (external), about the weight-bearing femur when toe-off takes place, which is more obvious for orthoses with shoe condition, continuing to opposite foot-strike when internal rotation begins again (3).

1.2. 2nd Data Collection: 22.03.2013

1.2.1. Temporal Distance Results ($X \pm \delta$)

		Barefoot	Orthoses+Shoe
Speed (m/s)		0.418	0.870
Stride Width (m)		0.204±0.020	0.160±0.035
Stride Length (m)		0.509±0.058	0.886±0.065
Cycle Time (s)		1.219±0.119	1.018±0.214
Step Length (m)	Left	0.273±0.051	0.425±0.030
	Right	0.236±0.032	0.461±0.057
Step Time (s)	Left	0.636±0.056	0.538±0.151
	Right	0.582±0.081	0.480±0.064
Stance Time (s)	Left	0.711±0.029	0.584±0.096
	Right	0.798±0.097	0.634±0.166
Swing Time (s)	Left	0.447±0.042	0.438±0.147
	Right	0.427±0.102	0.379±0.054
Stance Time / Cycle Time (%)	Left	≈ 58.3	≈ 57.4
	Right	≈ 65.5	≈ 62.3
Swing Time / Cycle Time (%)	Left	≈ 36.7	≈ 43.03
	Right	≈ 35.0	≈ 37.2
Double Limb Support Time (s)		0.338±0.102	0.198±0.049

Table 18 – Temporal Distance Results 2nd Data Collection

Regarding the **speed** values, it is possible to observe that orthoses with shoe present the double of speed value when compared to barefoot.

Stride width values are lower for orthoses with shoe than for barefoot, the same happening for **stride length**.

Cycle time presents higher values for barefoot condition than for orthoses with shoe condition, but values are closer for both conditions.

Observing the **step length** values, it is possible to observe that orthoses with shoe condition presents twice the values than barefoot condition, also **step time** is closer for those conditions, even that higher for barefoot than for orthoses with shoe condition.

Stance time is always higher than **swing time** (%), most of the situations in the proportion reported on literature.

Double Limb Support presents almost twice the values for barefoot condition when compared to orthoses with shoe condition.

Just like stated in the first data collection, this child presents worse performance when walking barefoot comparing to orthoses with shoe. Again it seems that the child feels more comfortable/fearless when walking with orthoses with shoes, and this can be due to the friction of shoe with the ground, the less kinesthetic information collected by the foot (since it does not contact with the ground) and as a consequence, the child could be more focused on walking forward.

1.2.2. Intersegmental angle on sagittal plane

1.2.2.1. Ankle

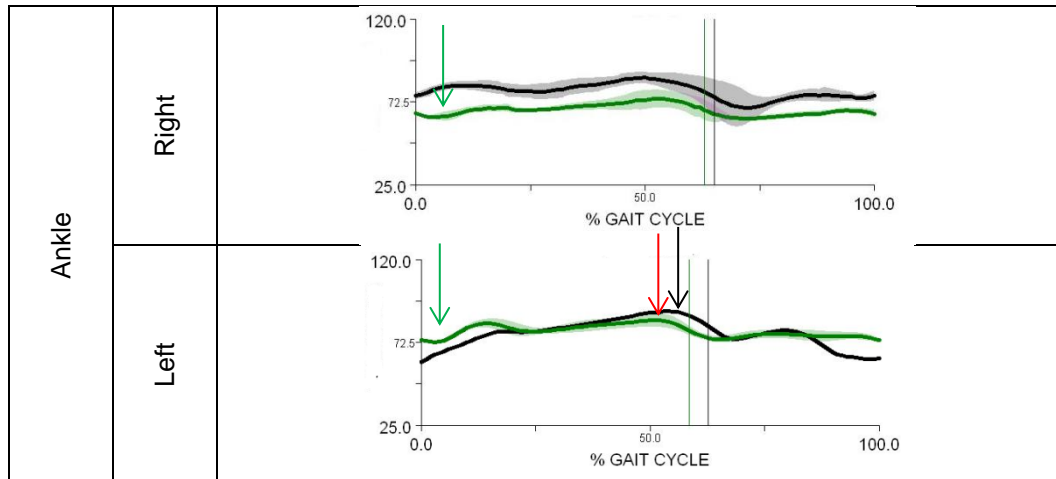


Table 19 – Ankle intersegmental angle on sagittal plane (dorsiflexion [+], plantar flexion [-]). Green – Orthoses with shoe; Black – Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.

After the contact of foot with the floor, plantar flexion is observed. That is possible to observe for the orthoses with shoe condition (green arrow) but not for the barefoot condition. That can be the result of changes made on the orthoses combined with a more controlled gait cycle expressed earlier in the temporal distance parameters for orthoses with shoe condition.

During unilateral stance phase, it is supposed to observe ankle moving in dorsiflexion until the maximum of 50% of gait cycle. Looking to the graphs, it is possible to observe that right ankle line is almost straight and left ankle presents a smooth peak, both being in dorsiflexion.

Orthoses were modified after the first data collection, giving more range of motion, being now possible to observe a peak of dorsiflexion, which is clear for left ankle in orthoses with shoe condition (red arrow), also barefoot condition has a dorsiflexion peak right after 50% of gait cycle (black arrow).

In the final phase of stance, plantar flexion occurs for all the conditions, being more obvious for barefoot.

After, ankle moves again in dorsiflexion, which occurs for both conditions.

Looking to the graphs and remembering the first evaluation, now orthoses with shoe condition have more range of motion between plantar and dorsi flexion. Although barefoot probably has less movement degree due to the new adaptations, the gait is also becoming more controlled and safe.

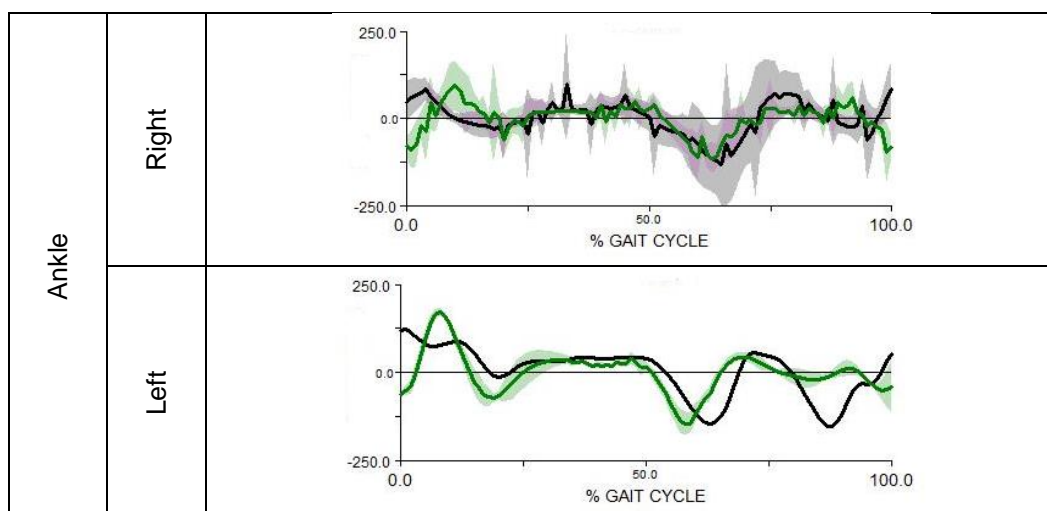


Table 20 – Ankle velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – Orthoses with shoe; Black – Barefoot.

It is expected to observe the lowest angular velocity on the ankle when compared to knee and hip.

Ankle presents maximum velocity right before the instant when the foot leaves the floor, approximately at 60% of gait cycle, as presented in the graphs. Is possible to observe, when foot touches the floor, that there is a brake and, at 60% of the cycle, acceleration happens for both feet, although left foot from barefoot condition presents another acceleration instant right after the 80% of the cycle.

1.2.2.2. Knee

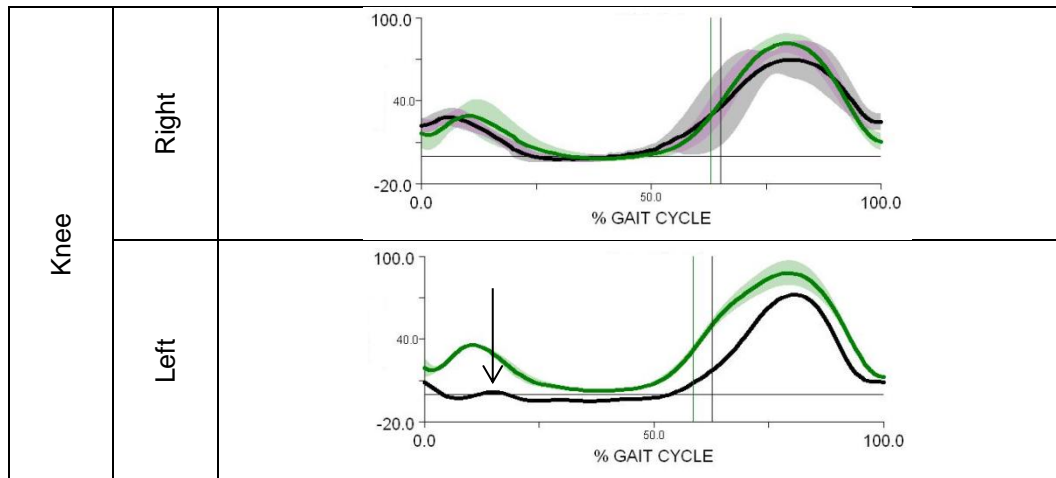


Table 21 - Knee intersegmental angle on sagittal plane (flexion [+], extension [-]).
 Green – Orthoses with shoe; Black - Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.

When heel strike occurs, knee should be in extension changing to flexion until the end of double support, and as is possible to observe for left knee of barefoot condition, it presents a smooth flexion peak later than expected (black arrow). Left knee presents this first flexion peak in a marked way when in orthoses with shoe condition.

At about 40% of gait cycle, an extension movement occurs, being possible to observe it clearly for orthoses with shoe condition and for right knee on barefoot condition.

After, knee returns to flexion, which occurs closer to the instant when foot leaves the floor and this movement presents a marked peak for all the conditions like expected for normal gait cycles.

Then knee returns to extension, finishing the gait cycle in total extension, as it is possible to observe on the graphs above.

This second data collection presents better movement between extension and flexion, especially for right barefoot condition, since in the

first data collection these movement variations were not so closer to the expected for normal gait. Knee under barefoot condition compensates the difficulties of ankle movement, and under orthoses with shoe condition, knee and ankle, are now doing their work better.

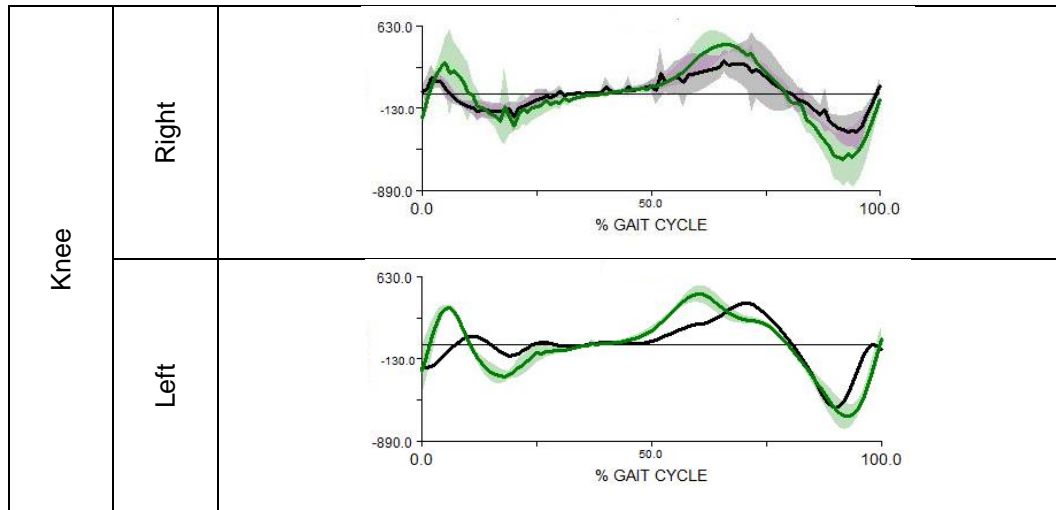


Table 22 - Knee velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – Orthoses with shoe; Black – Barefoot.

Knee has the highest angular velocity when compared to ankle and hip.

Knee presents maximum velocity when the foot leaves the floor and on the final phase of swing, approximately at 90% of gait cycle, as presented on the graphs and matching with the intersegmental angle graphs.

1.2.2.3. Hip

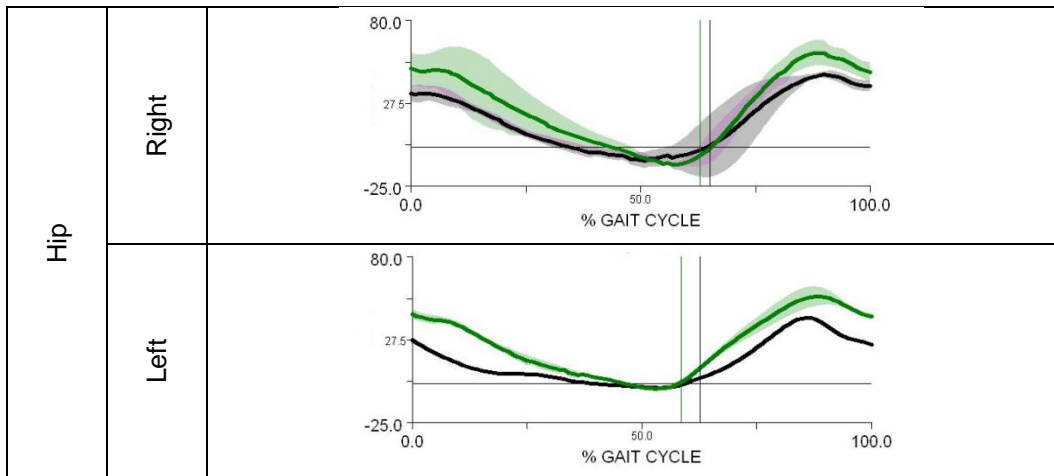


Table 23 - Hip intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – Orthoses with shoe; Black - Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.

When foot starts the contact with the floor, hip is on flexion, as presented on the graphs, for both conditions. Then, when stance's final phase occurs, it is possible to observe extension movement happening.

Graphs above show extension occurring for both conditions although a slighter curve for left hip on barefoot condition can be observed.

Hip returns to flexion right after, as presented on graphs for all conditions, although right hip needs more time to begin this return to flexion.

In a general way, hip movement is in agreement with the expected, some degrees of movement were lost when compared to the first data collection, especially for left hip.

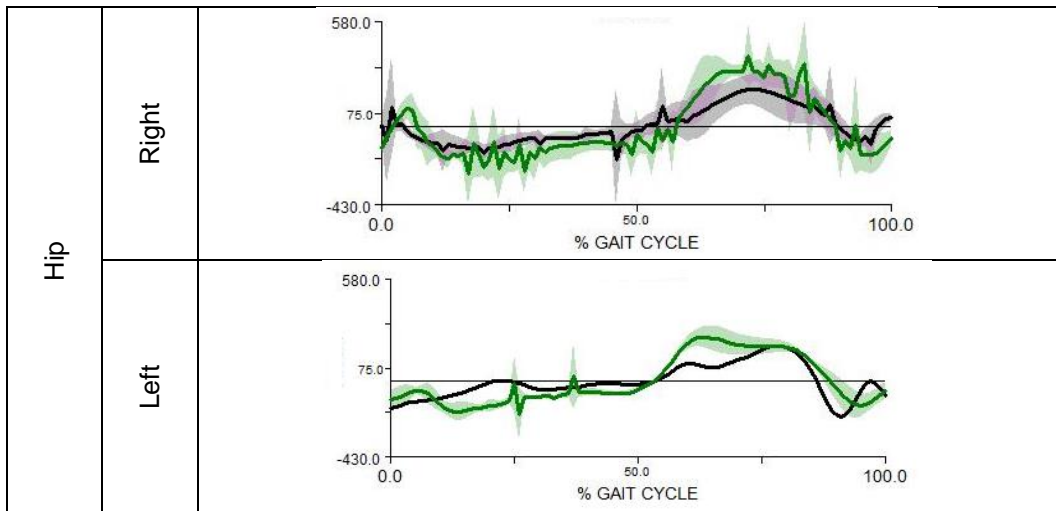


Table 24 - Hip velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – Orthoses with shoe; Black – Barefoot.

The hip's highest angular velocity should occur during the initial phase of swing, approximately at 70% of gait cycle, at the same time velocity at knee should be almost null, like presented on the graphs.

1.2.2.4. Pelvis

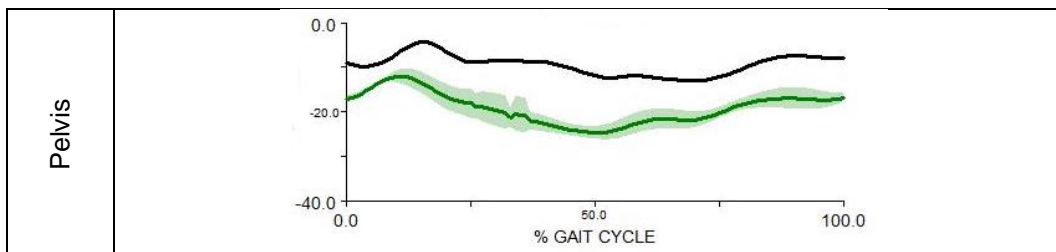


Table 25 - Pelvis intersegmental angle on sagittal plane (anterior flexion [+], posterior flexion [-]). Green – Orthoses with shoe; Black – Barefoot.

When analyzing pelvis on sagittal plane, pelvis tilt is being analyzed, meaning the movement between anterior and posterior flexion.

The pelvic movements, on this plane, consist in small variations in the degree of anterior pelvis tilt, which is inclined downwards anteriorly (3), although, just like on the first data collection, the child's pelvis is

posteriorly flexed, which is in agreement to the observed during the data collection.

It is desirable to obtain two peaks and two valleys. The valleys take place when toe-off and opposite toe-off happen, and peaks occur at late single stance and mid-to-late swing phase (3), although looking to the graphs that is not possible to observe. Comparing to the first data collection, the loss of those variations is observed, which is in agreement with the modifications detected for the other joints described on this plane.

1.2.3. Intersegmental angles on frontal plane

1.2.3.1. Ankle

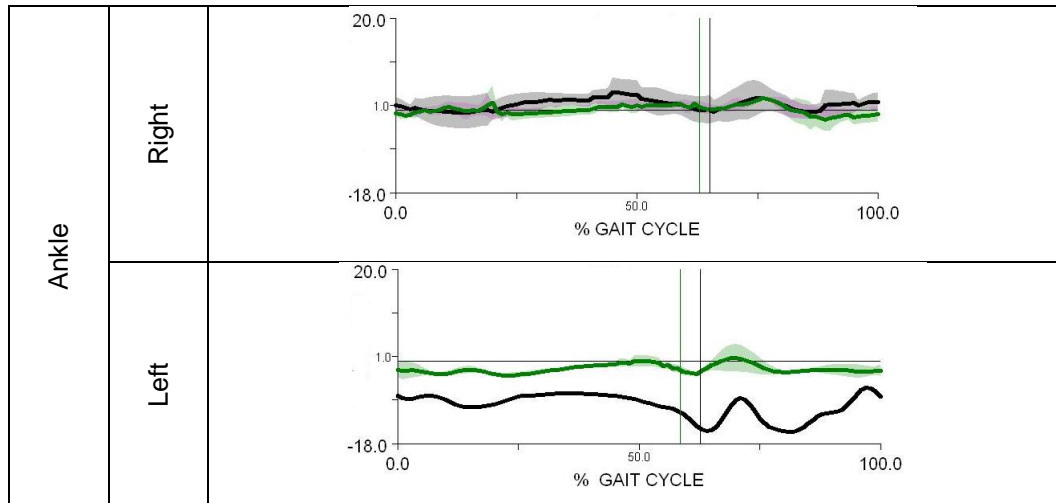


Table 26 - Ankle intersegmental angle on frontal plane (eversion [+], inversion [-]).
 Green – Orthoses with shoe; Black – Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.

It is important to remember that under static pose the child's feet are in eversion.

During the initial contact of foot with floor, ankle moves in eversion, in the positive way, which is not clear in most of the graphs presented above. Nevertheless it is possible to observe eversion's movement for left barefoot condition.

In the case of orthoses with shoe condition, their use will cause limitation on movement, especially on eversion and inversion. Although, it is observed that this condition increased degrees of movement when compared to first data collection, the same happening in the sagittal plane. Orthoses have suffered some modifications in order to allow more plantar and dorsiflexion range of motion.

It is also possible to observe on those graphs the inversion occurring too late in time and eversion occurring instead of inversion in the end of gait cycle, not forgetting that the feet tend to be on eversion.

During this second data collection, left foot presents inversion curve for barefoot and orthoses with shoe (this last one is better now comparing with first data collection).

Following, eversion should occur and ankle returns to neutral position, which is obvious for left barefoot, occurring smoothly to the rest of the conditions.

It is possible to observe that barefoot condition lost range of motion when comparing to the first data collection, which can be a sign of foot adaptation as a result of orthoses use, at the same time orthoses with shoe condition present small increase in the degrees of movement.

1.2.3.2. Knee

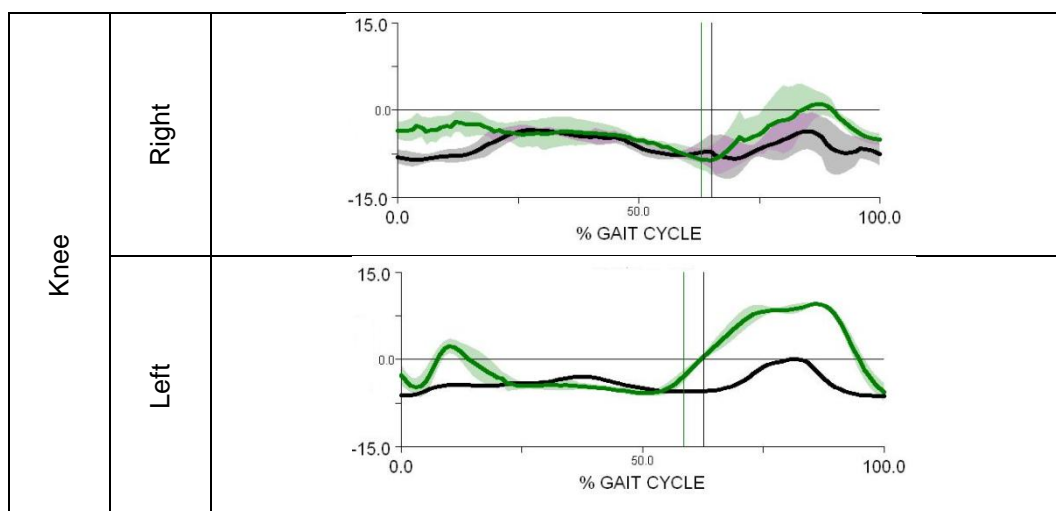


Table 27 - Knee intersegmental angle on frontal plane (adduction [+], abduction [-]).
 Green – Orthoses with shoe; Black – Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.

During the literature review about the child's disease, one of the characteristics referenced is the knee's *valgus* (1), which has influence

not only on the knee but also under the proximal joints that work in a chain with knee. Just as stated before, both feet, when in barefoot static pose, are in eversion.

In this plane, stability caused by joint restrictions is expected, although small movements occur between adduction and abduction.

During stance phase, it is supposed to observe an adducted knee increasing movement degree until about 60% of gait cycle. After that, it should return to abduction, swing phase, until the end of gait cycle.

Looking to the graphs, the movement described is possible to observe for left knee of orthoses with shoe condition.

At second data collection, the curves presented for barefoot and orthoses with shoe, even if not being equal to each other, present a similar pattern to the expected in regular cases, failing in the range of motion. Despite that, orthoses with shoe present an adduction peak higher than the expected in the beginning of the cycle.

It seems that the use of orthoses, according to the results presented on the graphs above, and especially for left knee, is helping the child to develop a movement pattern closer to the normal. Differences from the first data collection are observed.

1.2.3.3. Hip

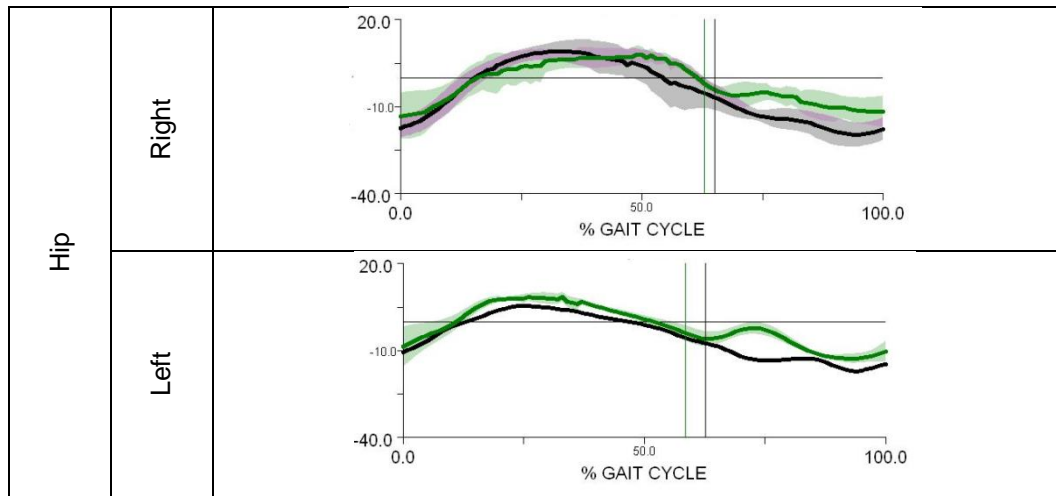


Table 28 - Hip intersegmental angle on frontal plane (abduction [+], adduction [-]). Green – Orthoses with shoe; Black - Barefoot. Vertical line represents the instant when foot leaves the contact with the floor.

During the initial contact, hip should change from 0° to about 6° of abduction at the end of double support phase. Looking to the graphs above, it is possible to observe that hip abductions increase, then slowly turns to adduction, which ends at approximately 60% of gait cycle, instant when hip should return to abduction, which is not observed.

It is possible to note, when comparing with first data collection, the lowest range of motion between adduction and abduction, and that both conditions are now closer.

1.2.3.4. Pelvis

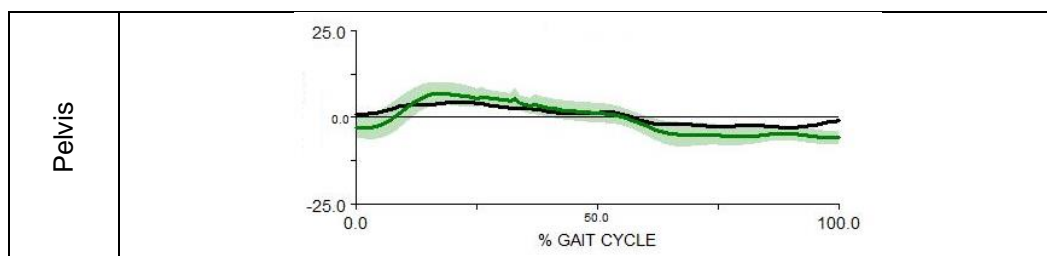


Table 29 – Pelvis intersegmental angle on frontal plane (Up [+], Down [-]). Green – Orthoses with shoe; Black – Barefoot.

Comparing with the first data collection, it is clear that range of motion was lost between evaluations.

In the frontal plane during early stance phase, the contralateral side of pelvis drops downward. The peak of pelvic obliquity occurs right after opposite toe-off, which corresponds to early stance phase on the weight-bearing limb (26) movement that is possible to observe, although with fewer range of motion than expected.

After that it drops, reaching a low point halfway through single stance, followed by a smaller peak elevation when opposite foot-strike occurs (3). After foot-strike, the ipsilateral pelvis drops to its lowest level at toe-off (3).

The loss of range of motion just as verified for sagittal plane can be due to the modifications promoted by the changes on the orthoses that add segment adaptations to the child's gait cycle or, according to the chapter visual impairment, correspond to one of the adaptations referred to this kind of population, which is a lower pelvis range of motion to induce safer walk (25).

1.2.4. Intersegmental angles in transverse plane

1.2.4.1. Ankle

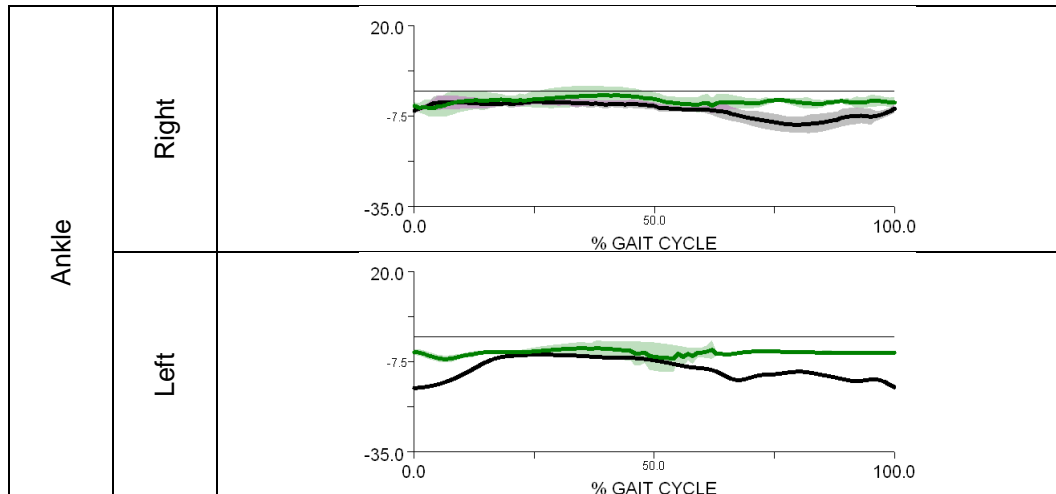


Table 30 - Ankle intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot.

In the beginning of initial contact, foot should be on neutral position or external rotation, followed by an internal rotation during the period in which foot establishes contact with the floor, which occurs during elevation of contralateral foot.

The use of orthoses will cause a limitation to those movements like explained earlier during frontal plane analysis (26).

Observing the graphs above, it is possible to observe that orthoses with shoe condition do not show significant movement degree changes, it goes like a straight line and barefoot presents a similar pattern, being worse now than in the first data collection when barefoot presented a curve closer to the normal gait.

However, it is important to note that now, also orthoses with shoe condition, for right ankle, is always on external rotation.

1.2.4.2. Knee

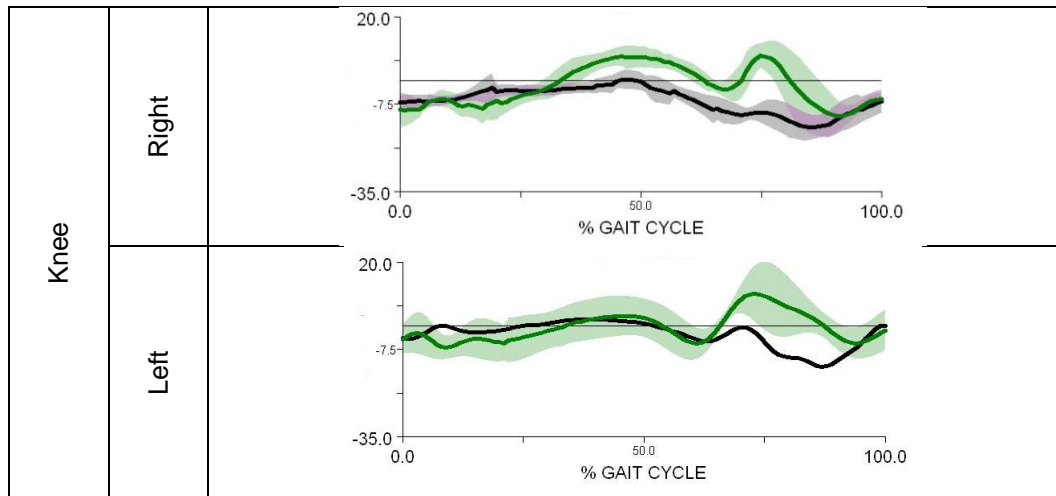


Table 31 - Knee intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black - Barefoot.

Knee rotation is linked to flexion and extension movement, so usually knee is in external rotation during gait cycle, being possible to observe internal rotation after foot strike. In the rest of the gait cycle, it is expected to observe external rotation increasing or decreasing the degrees of movement.

In the graphs above, knee is most of the time in external rotation, particularly for barefoot condition (both sides). Orthoses with shoe condition presents internal rotation movement, starting at above 30% of gait cycle for right knee and finishing right after the 75%. Left knee presents internal rotation after 50% of the gait cycle but it is not supposed to observe internal rotation in that phase of the cycle. The curve is closer to the presented on the normal gait cycle graphs.

Comparing to the first data collection, it is possible to observe positive modifications in the movement described by this joint.

1.2.4.3. Hip

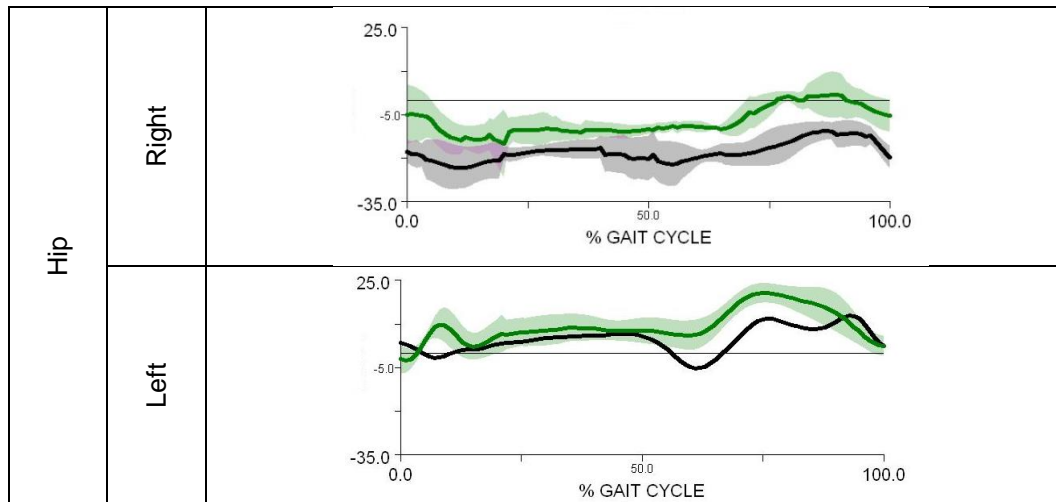


Table 32 - Hip intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot.

Those graphs show that for all the conditions the movement starts in external rotation except for left barefoot.

Then the hip should present an internal rotation movement.

Right hip for both conditions is almost always on external rotation changing the movement degrees along the gait cycle. It is possible to observe that orthoses with shoe condition at about 70% of the gait cycle starts a movement into internal rotation having a smooth internal rotation at about 85% of gait cycle.

Left hip for both conditions is almost always on internal rotation changing the degrees of movement along the gait cycle, being possible to observe that, after 50% of gait cycle, barefoot has an external rotation, and in the end of the cycle the internal rotation range of motion decreases almost into neutral position.

Comparing with first data collection differences are noticed, more obvious for right hip (now at internal rotation). The range of motion for right and left hip decreases.

1.2.4.4. Pelvis

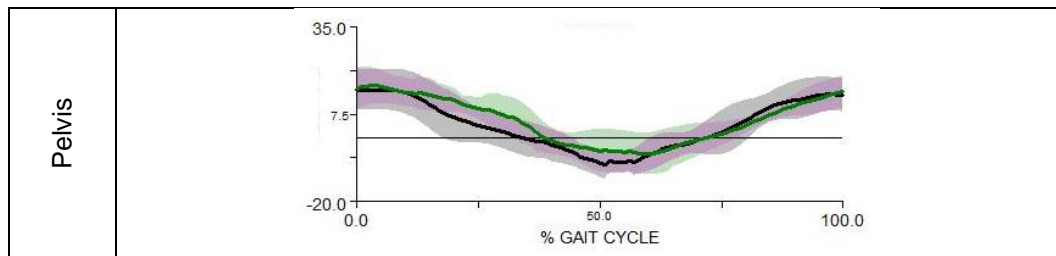


Table 33 - Pelvis intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – Orthoses with shoe; Black – Barefoot.

The peak of internal rotation occurs slightly after foot-strike (3) and pelvic rotation decreases in a faster way when heel strike occurs and is reversed as full weight is placed upon the foot (2), as is stated on the graphs above.

With load acceptance, the pelvis begins a counter-rotation (external), about the weight-bearing femur when toe-off takes place, continuing to opposite foot-strike when internal rotation begins again (3).

Compared to the first data collection, orthoses with shoe condition have lost range of motion especially in external rotation.

1.3. 3rd Data Collection: 19.07.2013

1.3.1. Temporal Distance Results ($X \pm \delta$):

		Barefoot	Orthoses+Shoe	Insole+Shoe
Speed (m/s)		0.253	0.425	0.535
Stride Width (m)		0.210 \pm 0.024	0.213 \pm 0.018	0.204 \pm 0.034
Stride Length (m)		0.348 \pm 0.138	0.585 \pm 0.076	0.603 \pm 0.122
Cycle Time (s)		1.374 \pm 0.500	1.376 \pm 0.095	1.127 \pm 0.136
Step Length (m)	Left	0.236 \pm 0.042	0.284 \pm 0.044	0.315 \pm 0.059
	Right	0.114 \pm 0.129	0.300 \pm 0.046	0.291 \pm 0.082
Step Time (s)	Left	0.743 \pm 0.116	0.671 \pm 0.056	0.577 \pm 0.077
	Right	0.751 \pm 0.151	0.703 \pm 0.078	0.542 \pm 0.074
Stance Time (s)	Left	1.058 \pm 0.169	0.895 \pm 0.100	0.657 \pm 0.154
	Right	1.025 \pm 0.383	0.888 \pm 0.097	0.722 \pm 0.095
Swing Time (s)	Left	0.525 \pm 0.066	0.475 \pm 0.075	0.438 \pm 0.064
	Right	0.399 \pm 0.155	0.474 \pm 0.031	0.432 \pm 0.042
Stance Time / Cycle Time (%)	Left	\approx 77	\approx 65.04	\approx 58.30
	Right	\approx 74.6	\approx 64.53	\approx 64.06
Swing Time / Cycle Time (%)	Left	\approx 38.21	\approx 34.52	\approx 38.86
	Right	\approx 29.04	\approx 34.45	\approx 38.33
Double Limb Support Time (s)		0.5557 \pm 0.170	0.419 \pm 0.132	0.246 \pm 0.132

Table 34 – Temporal Distance Results 3rd Data Collection

Regarding the **speed** values, it is possible to check that between conditions speed increases. Barefoot has the lowest speed and, the most recent condition, insole with shoe, the highest speed value.

Stride width values are lower for insole with shoe condition. The other two conditions have closer values, although the stride width values are higher for orthoses with shoe condition.

With insole with shoe condition, the child presents a higher **stride length** compared to the other two conditions tested during the third data collection.

Orthoses with shoe condition present higher **cycle time** value than barefoot, and insole with shoe condition presents the lowest value for cycle time. However, the difference between barefoot and orthoses with

shoe is very small and stance and swing phases do not take equal time for both conditions.

Observing the **step length** values related to the three conditions, it is possible to observe that step length is lower for barefoot condition than to the other two conditions, being higher for the condition insole with shoe. It is possible to observe that right step length takes lower values than left step length, with the exception of orthoses with shoe condition.

For **step time** it is possible to observe higher values for barefoot when compared with the two other conditions and insole with shoe presents the lowest values. Also right step time presents higher values than left step time in two of the conditions, barefoot and orthoses with shoe.

Stance time is always higher than **swing time** (%), but even when orthoses with shoe and insole with shoe are closer to the expected, meaning a 60% to 40% relation, for barefoot condition in this data collection the relation is above 70% to 30%.

Double Limb Support is expressed by higher values on barefoot condition when compared to the others.

Like on the other collections, barefoot seems to be the worst condition and the child presents better performance when using foot orthoses. Between the two orthoses in evaluation, in this data collection, even when not using it for a long time (a month), insole with shoe condition seems to have better results than the other condition. It is known that insole has a proprioceptive effect under the foot, as stated earlier (3), which can contribute to those results.

1.3.2. Intersegmental angle on sagittal plane

1.3.2.1. Ankle

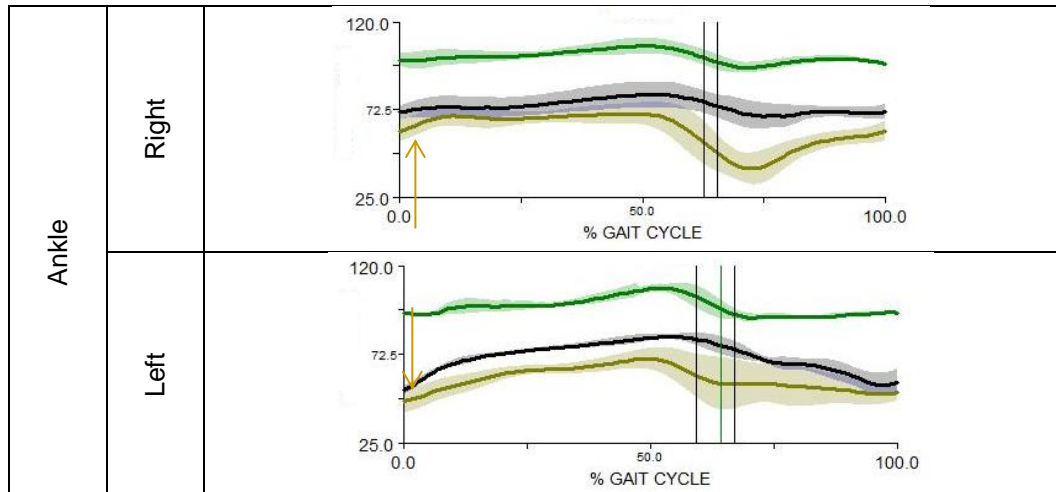


Table 35 - Ankle intersegmental angle on sagittal plane (dorsiflexion [+], plantar flexion [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.

In the beginning of gait cycle, after the contact of foot with the floor, it is expected to observe plantar flexion. Looking to the graphs above, it is possible to observe this movement for insole with shoe condition (beige arrow).

Then, during unilateral stance phase, it is observed that ankle moves in dorsiflexion until the maximum of 50% of gait cycle, although not so much as expected. Smooth dorsiflexion curves for orthoses with shoe and for barefoot conditions can be observed. In case of right foot, the curve for barefoot condition seems very similar to orthoses with shoe condition curve.

Both conditions, barefoot and orthoses with shoe have smaller peaks, being more linear, when compared with insole with shoe condition. This means less switch between dorsi and plantar flexion.

Insole with shoe presents those movements more clearly, dorsi and plantar flexion, at approximately 50% of gait cycle.

In the final phase of stance, a new plantar flexion should occur very clearly for all conditions, easy to identify for insole with shoe.

After, ankle returns to a dorsiflexion movement, which is clear in almost all conditions evaluated except for left barefoot conditions from which it is possible to verify that the curve decreases to plantar flexion, instead of dorsiflexion.

It seems that insole with shoe condition is promoting range of motion on ankle, specially for right ankle. Barefoot continues losing movement between dorsi and plantar flexion when compared to the earlier evaluations.

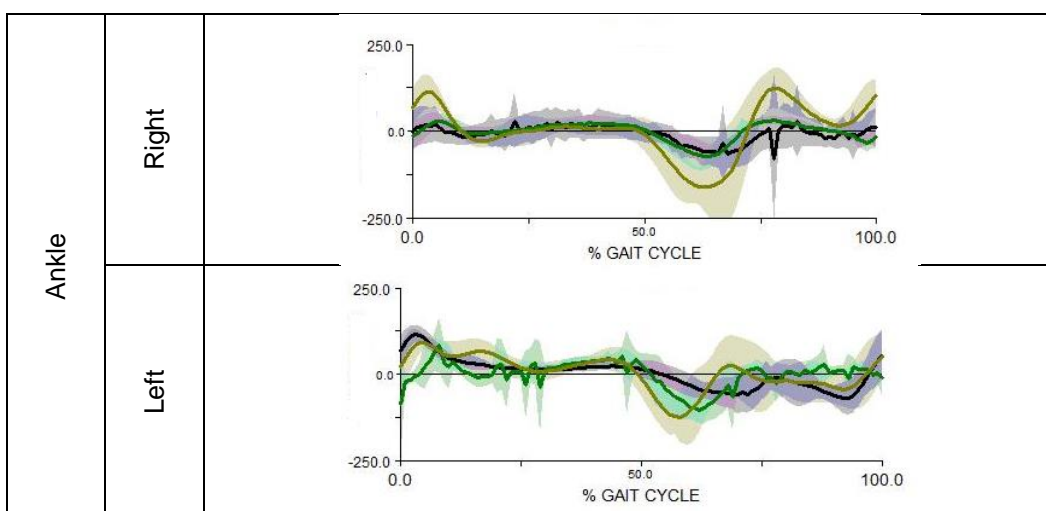


Table 36 – Ankle velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

It is expected to observe the lowest angular velocity on the ankle when compared to knee and hip.

Ankle presents maximum velocity right before the instant when the foot leaves the floor, approximately at 60% of gait cycle, as presented in the graphs.

Is easy to observe when looking to right foot graphs, in the beginning of gait cycle, when the foot touches the floor, that a braking moment occurs and, at 60% of the cycle, for both feet, an acceleration moment is possible to observe. However left foot acceleration is lower than the expected.

1.3.2.2. Knee

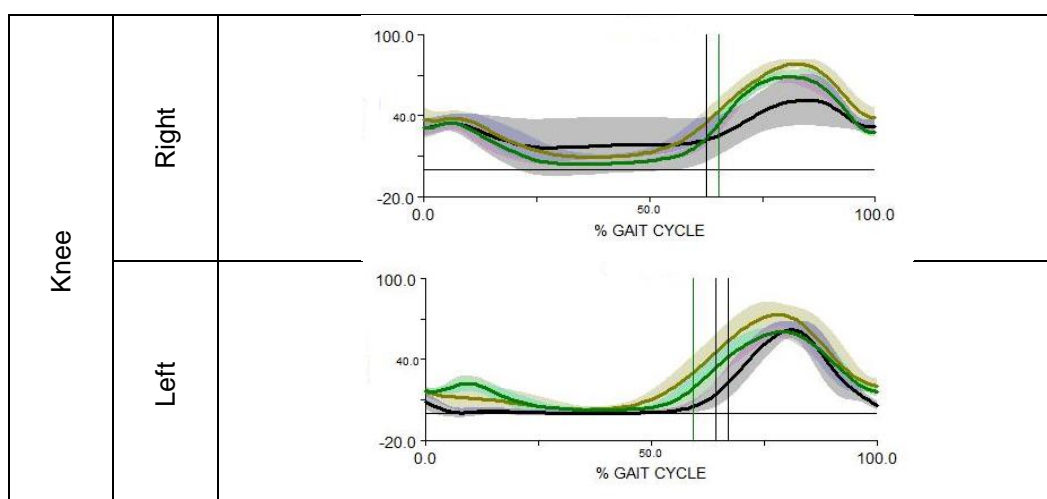


Table 37 - Knee intersegmental angle on sagittal plane (flexion [+], extension [-]).
 Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.

When heel strike occurs, knee should be in extension returning to flexion until the end of double support. Looking to orthoses with shoe condition, it is possible to observe it, being the closest one to the expected in normal situations, although left knee presents a smooth flexion peak a little later than the expected, and for right knee (the same condition) a smaller flexion curve is observed.

It is also expected an extension movement until about 40% of gait cycle, which again is clear for orthoses with shoe condition. Right and left knee on insole with shoe condition also present it, although left knee presents less range of motion.

After, knee returns to flexion that occurs closer to the instant when foot leaves the floor. In the graphs above, for all conditions, it is possible to observe it, even if in some cases it happens earlier. It is possible to observe one exception for right knee on barefoot condition in which flexion peak is smaller.

After this flexion peak, knee returns to extension, and finishes the gait cycle in extension, which is observed in all graphs, although more obvious in some.

Between second data collection and the present data collection, insole with shoe condition was added. It seems that in terms of knee movement, a good adaptation was achieved, especially for flexion at toe-off. Orthoses with shoe movement is almost equal to the last data collection, but barefoot condition, especially right barefoot (that was the best one in the last evaluation) seems to lose some degrees of movement, which can result from the adaptation period to the new condition (insole).

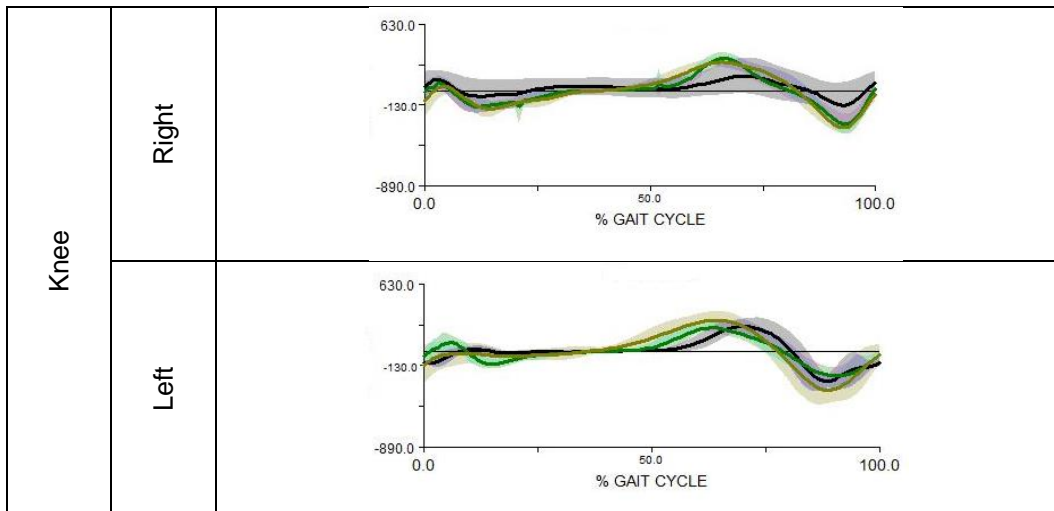


Table 38 - Knee velocity on sagittal plane (velocity increase [+], velocity decrease [-]).
 Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

It is expected to observe the highest angular velocity on the knee when compared to ankle and hip.

Knee presents maximum velocity when the foot leaves the floor and on the final phase of swing, approximately at 90% of gait cycle, like presented in the graphs above.

1.3.2.3. Hip

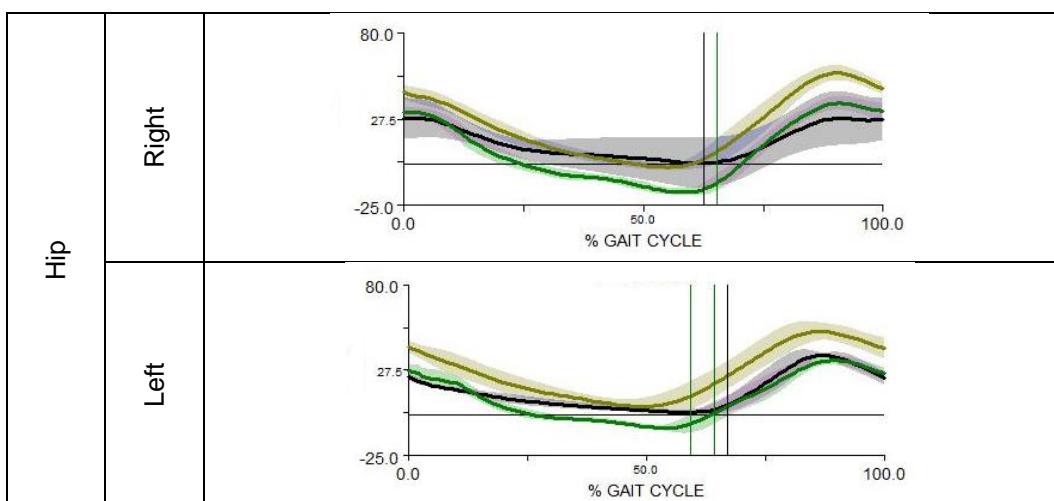


Table 39 - Hip intersegmental angle on sagittal plane (flexion [+], extension [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.

At initial contact, when foot starts the contact with the floor, hip should be on flexion, like presented in all graphs for all conditions.

After, when it reaches the final phase of stance, extension should occur.

In the graphs above, it is possible to see extension occurring for all conditions, although smoothly during barefoot condition.

After, hip returns to flexion, which is obvious in all graphs, although right hip needs more time to return to flexion.

In a general way, hip movement is in agreement with the expected. Comparing the three conditions, barefoot is the one with less range of motion between flexion and extension, and orthoses have increased extension degrees, when compared with the other data collections.

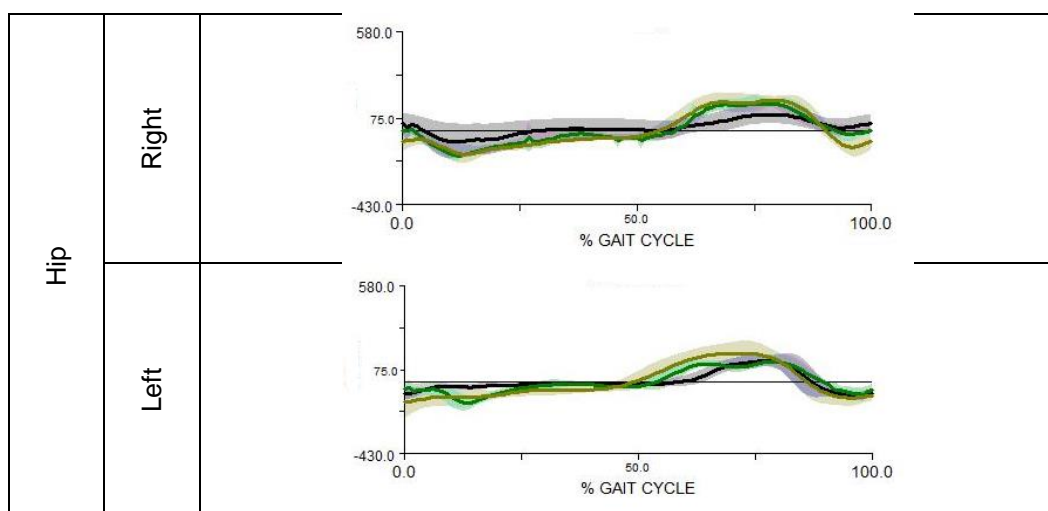


Table 40 - Hip velocity on sagittal plane (velocity increase [+], velocity decrease [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

It is expected to observe the highest angular velocity of the hip during the initial phase of swing, approximately at 70% of gait cycle. At the same time, velocity at knee should be almost null, as presented in the graphs above.

1.3.2.4. Pelvis

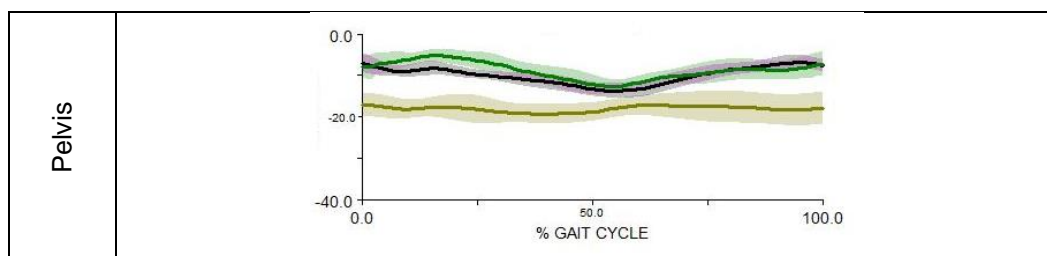


Table 41 - Pelvis intersegmental angle on sagittal plane (anterior flexion [+], posterior flexion [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

When analyzing pelvis on sagittal plane, pelvis tilt is being analyzed, meaning the movement between anterior and posterior flexion.

Like on the other two data collections made before, pelvis is not on anterior flexion but in posterior flexion. The degree of movement in the graphs above is different from the expected and presented on literature review, since the two peaks and the two valleys are not presented.

For insole with shoe data collection, the graphs present an almost straight line.

The loss of range of motion can result from all the modifications promoted in the lower limb but are also described on the literature reviewed on the visual impairment chapter as a characteristic of low vision adaptations in order to avoid the falls and turn walking safer (25).

1.3.3. Intersegmental angles on frontal plane

1.3.3.1. Ankle

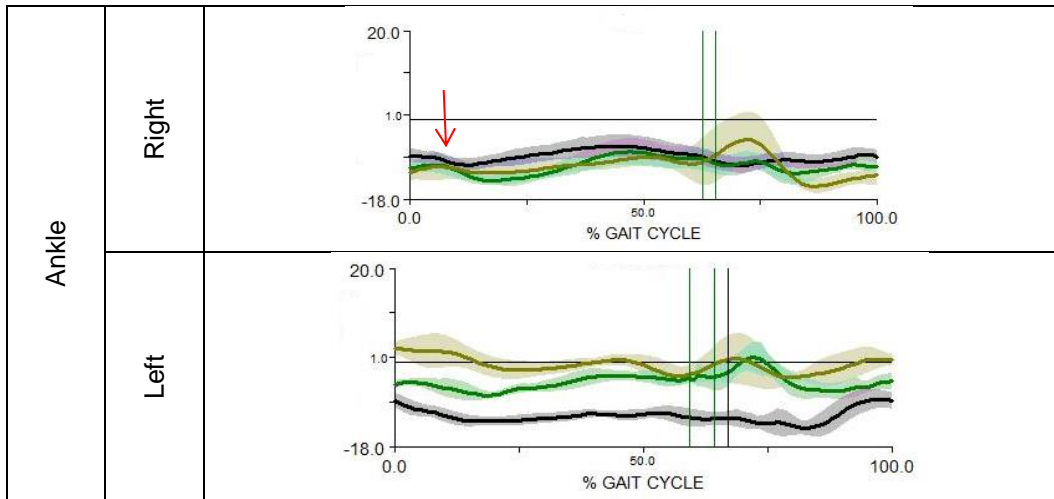


Table 42 - Ankle intersegmental angle on frontal plane (eversion [+], inversion [-]).
Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.

Under static pose (barefoot) the child tends to have feet on eversion position.

During the initial contact of foot with floor, ankle should be in eversion, which is not clear in all the graphs. However, it is possible to observe the eversion peak in some data (red arrow), for example for conditions: right orthoses with shoe and right insole with shoe.

According to the literature, the use of orthoses will cause limitation on eversion and inversion movement (26), although what is seen from data collection to data collection is that this condition is achieving more degrees of movement.

It is possible to observe that insole with shoe condition presents inversion curve for right ankle, but the other two conditions do not. In

addition, left ankle (the same condition) presents inversion too early, presenting eversion earlier too. Barefoot does not present inversion.

Following the return to neutral position should occur, being possible to observe eversion peak for insole with shoe condition, for orthoses with shoe conditions, and left barefoot.

Barefoot seems now worse, once more losing degrees of movement, which can mean that it is readapting to the conditions to which it is being submitted: orthoses and insole.

1.3.3.2. Knee

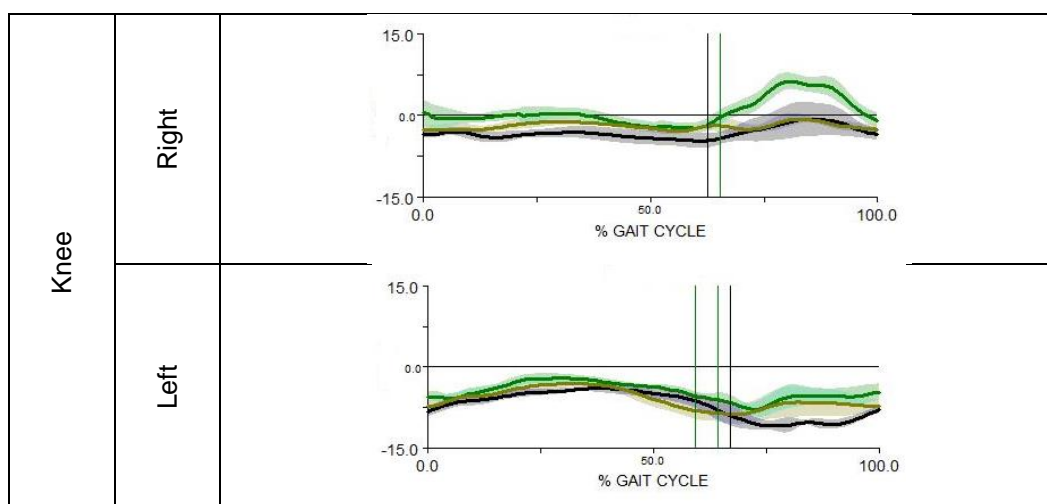


Table 43 - Knee intersegmental angle on frontal plane (adduction [+], abduction [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.

During the literature review about the child's disease, one of the characteristics referenced is the knee's *valgus* (1), which has influence not only on the knee but also under the proximal joints that work in a chain with knee, just like stated before both feet, when in barefoot static position, are in eversion.

It is supposed to observe a pattern of stability because of joint restrictions, although small movements occur between adduction and abduction.

During stance phase, it is supposed to observe the knee on adduction, increasing the degrees until more or less 60% of gait cycle, and after, during the swing phase, the knee changes to abduction, until the end of gait cycle.

On orthoses with shoe condition, even having a curve closer to the expected for right knee, does not have it for left knee. Right knee is better now and left knee is worse.

It is possible to observe that only right knee presents a curve closer to the expected. The other conditions do not correspond to the expected. Also, for insole with shoe condition, results are far away from the expected on regular situations.

That can be a consequence of the insole's use, first because the child now uses insoles instead of orthoses almost all the time, and he started to use them only one month before the evaluation, being possible that the child is not well adapted yet to this new condition.

1.3.3.3. Hip

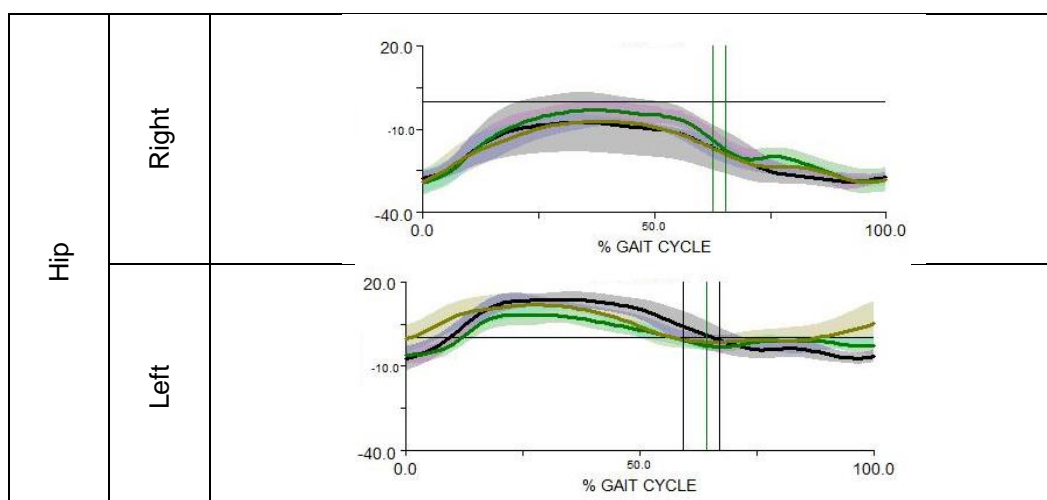


Table 44- Hip intersegmental angle on frontal plane (abduction [+], adduction [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe. Vertical line represents the instant when foot leaves the contact with the floor.

During the initial contact, hip should be with 0° changing to a 6° abduction at the end of double support phase, which is not what graphs show, even when insole with shoe condition for left hip starts at 0° . In every graph it is possible to observe that a small increase on hip abduction occurs although right hip graphs show it always under adduction.

Then hip should move slowly into adduction finishing it at about 60% of gait cycle, returning to abduction, which is possible to observe, even when it is not to clear for all conditions, and better for insole with shoe for left hip.

Third data collection presents a lower range of motion between adduction and abduction.

1.3.3.4. Pelvis

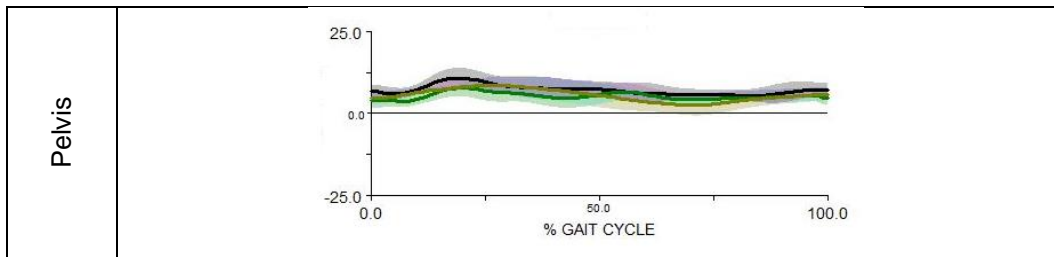


Table 45 – Pelvis intersegmental angle on frontal plane (Up [+], Down [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

Comparing this data collections graph with the other two data collections made before, the loss of range of motion is obvious, the lines are almost straight, only the first peak is possible to observe, which corresponds to the peak of pelvic obliquity that occurs right after opposite toe-off in the early stance phase on the weight-bearing limb.

The lost of range of motion as verified for sagittal plane can be due to the modifications promoted by the changes on the orthoses, that add segments adaptations to the child's gait cycle or to one of the adaptations referred to this kind of population, which is a lower pelvis range of motion in order to walk safely (25).

1.3.4. Intersegmental angles in transverse plane

1.3.4.1. Ankle

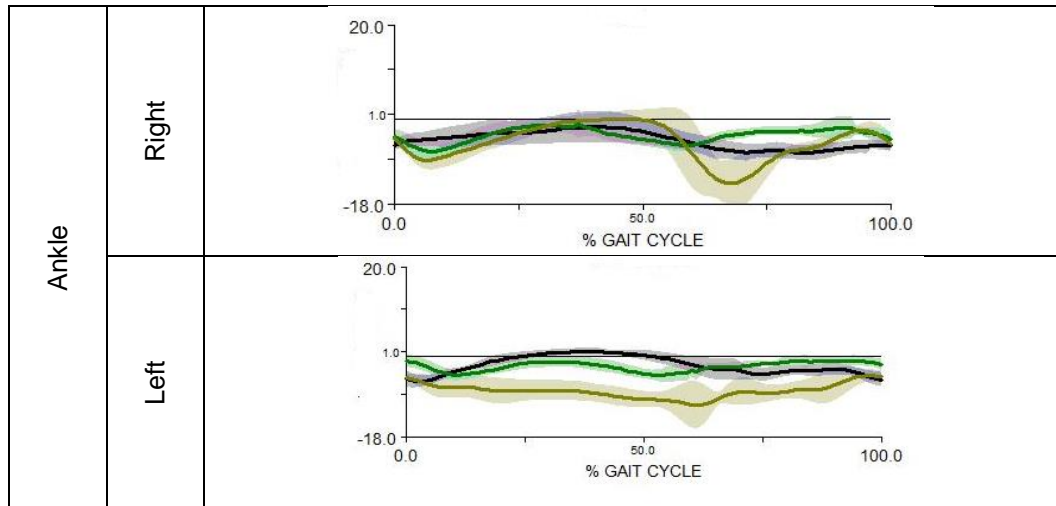


Table 46 - Ankle intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

In the beginning of initial contact with the floor, foot should be in neutral position or external rotation, followed by an internal rotation during the period that foot establishes in the floor, which occurs during elevation of contralateral foot.

The use of orthoses will cause movement limitation (26), so observing the graphs it is possible to conclude that for this condition the movement is described by an almost straight line, although comparing with the second data collection some movement degree was achieved.

Like observed during the last data collection, barefoot condition has lost range of motion.

Insole with shoe condition presents for right foot a curve similar to the expected for normal gait cycle although left foot even when standing in external rotation does not present significant range of motion.

1.3.4.2. Knee

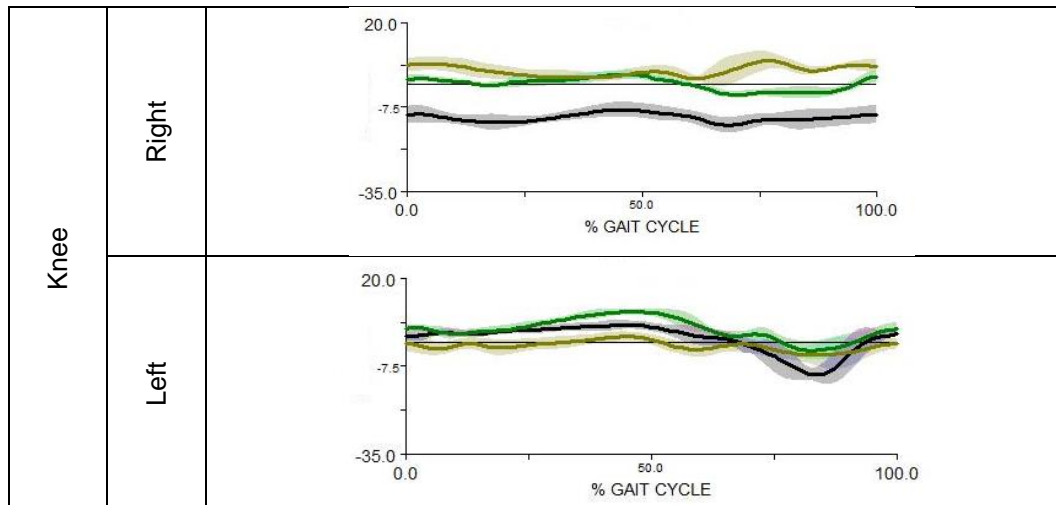


Table 47 - Knee intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

Knee rotation is linked to flexion and extension movement, so usually knee is in external rotation during gait cycle, only modifying the amplitude range along that, being although possible to observe an internal moment after foot strike.

However, what graphs show is not in agreement with that, being possible to observe different patterns for each of the knees.

In a general way, right knee seems to lose range of motion in this last evaluation, when comparing to the other two evaluations done before. Right knee in insole with shoe condition does not present considerable range of motion presenting a line that is almost straight and always in internal rotation. The same happens with orthoses with shoe condition, although for this condition there is a change between internal and external rotation in the middle of gait cycle. Barefoot condition is on external rotation but also in a straight line with few movement.

Left knee for orthoses with shoe condition presents a similar pattern to the presented for right knee, starting with internal rotation and changing to external rotation. For barefoot, it is almost described as a straight line, starting with an internal rotation that changes to external rotation with a peak for external rotation at about 80% of gait cycle. Left insole with shoe is usually on external rotation being described by a straight line too.

Comparing with previous evaluations, it seems that knee under transverse plane is losing range of motion.

1.3.4.3. Hip

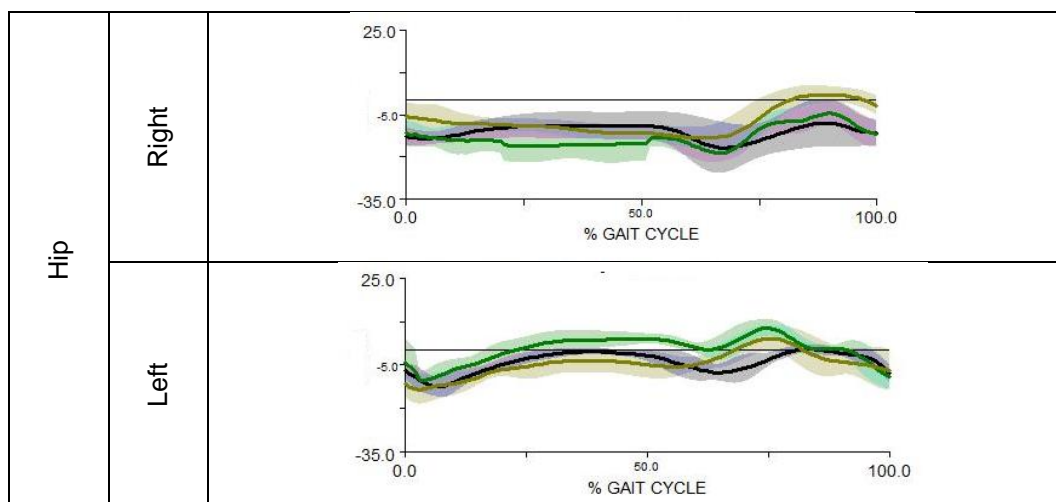


Table 48 - Hip intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – orthoses with shoe; Black – Barefoot; Beige – insole with shoe.

The movement starts with external rotation for all the conditions and, following the initial contact, it is expected to observe the hip rotating internally and returning to external rotation at the end of gait cycle.

Despite the changes in the range of motion of each condition, barefoot is always at external rotation for left and right hip, even when left hip shows two instants in neutral position (when black curve touches the neutral line).

Left orthoses with shoe condition curve presents internal rotation between 25% and 75% of gait cycle, which is the closest to normal gait cycle observed during this evaluation, but right orthoses with shoe condition is always on external rotation.

Left and right insole with shoe condition is almost all the gait cycle on external rotation, both with two instants of internal rotation, right hip starts at about 75% of gait cycle and goes until the end of the gait cycle, and left hip has a peak at 75% of gait cycle returning to external rotation.

1.3.4.4. Pelvis

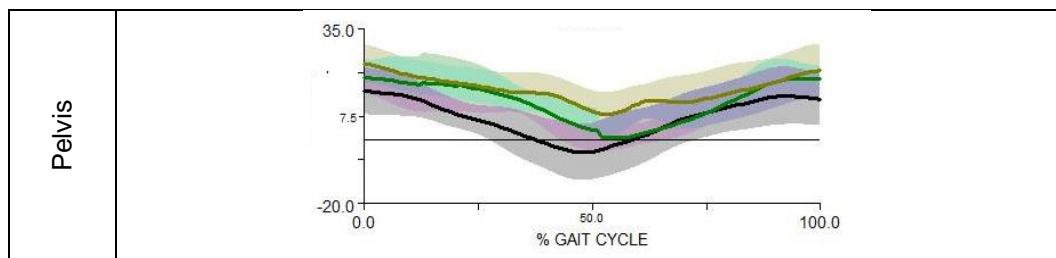


Table 49 - Pelvis intersegmental angle on transverse plane (internal rotation [+], external rotation [-]). Green – orthoses with shoe; Black - Barefoot; Beige – insole with shoe.

The peak of internal rotation occurs slightly after foot-strike (3) and pelvic rotation decreases in a faster way when heel strike occurs and is reversed as full weight is placed upon the foot (2), as stated on the graphs above.

With load acceptance, the pelvis begins a counter-rotation (external), about the weight-bearing femur when toe-off takes place, continuing to opposite foot-strike when internal rotation begins again (3).

Compared to the first two data collections, orthoses with shoe have lost range of motion. In these data collection it does not do external rotation.

Insole with shoe condition has also a small range of motion when compared to the expected for a regular gait cycle and with the other two conditions presented.

1.4. Barefoot and Orthoses with shoe conditions along time

Even though all the conditions were already described, it is also interesting to observe if there were, along the time, modifications on the two conditions always evaluated, since those conditions were not directly modified.

1.4.1. Barefoot Condition

1.4.1.1. Temporal Distance Results ($X \pm \delta$)

		1 st Data Collection	2 nd Data Collection	3 rd Data Collection
Speed (m/s)		0.804	0.418	0.253
Stride Width (m)		0.088±0.010	0.204±0.020	0.210±0.024
Stride Length (m)		0.809±0.043	0.509±0.058	0.348±0.138
Cycle Time (s)		1.007±0.142	1.219±0.119	1.374±0.500
Step Length (m)	Left	0.377±0.0057	0.273±0.051	0.236±0.042
	Right	0.435±0.047	0.236±0.032	0.114±0.129
Step Time (s)	Left	0.490±0.086	0.636±0.056	0.743±0.116
	Right	0.516±0.066	0.582±0.081	0.751±0.151
Stance Time (s)	Left	0.607±0.063	0.711±0.029	1.058±0.169
	Right	0.600±0.129	0.798±0.097	1.025±0.383
Swing Time (s)	Left	0.382±0.071	0.447±0.042	0.525±0.066
	Right	0.426±0.039	0.427±0.102	0.399±0.155
Stance Time / Cycle Time (%)	Left	≈ 60.3	≈ 58.8	≈ 77
	Right	≈ 59.6	≈ 65.5	≈ 74.6
Swing Time / Cycle Time (%)	Left	≈ 37.9	≈ 36.7	≈ 38,21
	Right	≈ 42.3	≈ 35.0	≈ 29.04
Double Limb Support Time (s)		0.203±0.091	0.338±0.102	0.557±0.170

Table 50 – Temporal Distance Results for Barefoot Condition

Along the time, barefoot condition presents differences in the temporal and spatial parameters.

It is possible to observe **speed** decreasing along time, which means a higher control over gait, which will influence the other parameters.

Stride width decreases significantly between the first and the second evaluation but has a small increase from second evaluation to the third one.

Stride length also decreases along the time for less than a half when comparing the first and the third evaluations.

Cycle time has an increase along the time and the same happens with **step time**.

Step length also decreases along the time, being more obvious for right foot than for left foot when comparing the results obtained in the third evaluation.

Stance time and **swing time** become closer to the normal, which is 60% / 40%, along the time, with one exception for the right foot in the last evaluation. The relation of time of stance and swing during a gait cycle shows that stance takes more time than swing along the evaluations, but in the last evaluation the data obtained show that an increment on stance time happens, and it is exceeding the expected.

Double support time also increases along time.

What is observed along the time and that justifies the evolution that this child has in barefoot condition is a more mature and safe gait. In the first evaluation, as stated before, it was necessary to help the child walking because usually he falls, his parents reported that at home sometimes he prefers to crawl instead of walk avoiding the falls. On the second and third evaluation, he did not need help and that happens in result of a more mature and safe gait, resulting specially in higher periods of single and double support but also swing takes more time in the third evaluation that in the first one. Looking at the table results above, it is possible to check it.

1.4.1.2. Intersegmental Angles on Sagittal Plane

1.4.1.2.1. Ankle

When observing ankle graphs (Table 3) during the first evaluation, it is possible to observe that barefoot presents similar movement pattern for right and left ankle on sagittal plane.

The initial movement of plantar flexion is not observed, but a smooth movement of dorsiflexion until 50% of gait cycle is possible to observe followed by a plantar flexion movement, although it does not achieve huge movement degree.

Then ankle returns to dorsiflexion and it should return to plantar flexion in the end of gait cycle, which is not evident.

Second data collection graphs (Table 19) show some differences between right and left ankles under barefoot movement condition.

The huge difference is the dorsiflexion movement for left ankle, which happens closer to the 50% of gait cycle.

Third data collection graphs (Table 35) show smooth curves for barefoot, meaning that movement between dorsiflexion and plantar flexion is almost absent.

The dorsiflexion movement that should occur after is possible to observe although not too clear.

After, ankle moves again in dorsiflexion, which is possible to observe, even though smoothly, for right foot, but for left barefoot it is possible to check that plantar flexion occurs instead of dorsiflexion.

Between the first and the third evaluation, some differences are observed. The first difference noted is the loss of range of motion between plantar and dorsiflexion, which is more obvious between first and second evaluation. Those differences could be the result of the adaptations required by the orthoses modifications (between 1st and 2nd data collection) and the introduction of insole (between 2nd and 3rd data collection). Another reason to explain the differences is the fact described on visual impairment chapter that people with low vision usually strike the floor with the entire foot (18) (19) (24) in order to collect more kinesthetic information that helps with gait control.

1.4.1.2.2. Knee

On sagittal plane, knee graphs (Table 5 and 21) show between first and second data collection a better graph of movement (meaning that it is taking the waves expected for normal gait cycle) even when the first flexion peak is still not obvious specially for left knee, right knee presents it, the same happening on third data collection.

It is clear that between first and second data collection there are improvements in the movement graphs, with curves and peaks closer to the expected for regular gait, although between second and third (Table 37) data collection, especially right knee loses movement degrees.

Extension is observed for both knees, like expected in the end of the cycle, during the first evaluation but on the second evaluation only right knee presents it. During third evaluation, it is also possible to observe extension movement for both knees taking place until the end of gait cycle, almost achieving neutral position for left knee.

The last evaluation, the third one, seems to present a movement pattern closer to regular ones when compared with the two other evaluations.

It seems that the orthoses modifications done after first data collection brought new and better achievements for knee movement under barefoot condition, but insole with shoe required new adaptations especially for barefoot condition.

1.4.1.2.3. Hip

Hip graphs, during the first evaluation (Table 7), present a pattern of movement closer to the expected and the same happens on second data collection (Table 23), although left hip curve is smoother for left hip than for right hip.

Third evaluation (Table 39) presents a pattern similar for both hips and this pattern is closer to the expected for regular ones.

In a general way hip does not present significant changes if compared to a normal gait cycle.

1.4.1.2.4. Pelvis

Along the time its observed that pelvis is positioned backward instead of forward as stated on literature (3).

During the first evaluation (Table 9), when observing pelvis on the sagittal plane it is possible to verify that its movement graph is not in agreement with the expected for regular gait, the two peaks and the two valleys were replaced by variations on the curve.

Along the time the graphs (Table 25 and Table 41) that describe pelvis movement under sagittal plane continue not presenting the two valleys and the two peaks and at the same time losing range of motion. That, as written before, can be the result of the adaptations promoted along

the time on the lower limb, but can also represent a characteristic of low vision, described on the visual impairment chapter, to improve safety to the gait (25).

1.4.1.3. Intersegmental Angles on Frontal Plane

1.4.1.3.1. Ankle

During the first evaluation (Table 10), frontal plane graphs show the absence of initial eversion peak for left ankle presenting it for right ankle. Following when inversion movement occurs, at about 50% of gait cycle, right foot presents it more clearly than left foot. In order to return to neutral position, an eversion movement occurs, being more clearly for left ankle than for right ankle.

Second data collection graphs (Table 26) show in a general way that ankle loses degrees of movement under this plane. However, even when right ankle decreases movement degree, between inversion and eversion, left ankle increases movement degree, when compared to the first evaluation.

Third data collection graphs (Table 42) show again that barefoot is losing degrees of movement in this plane of movement and that ankle seems to remain in inversion. Although, right ankle shows an eversion movement right after the contact of foot with the floor returning to inversion. After that, eversion occurs at the end of the cycle, more obvious for left foot.

It is possible to observe along the data collections that feet are losing range of motion in this plane, which can be the result of orthoses and insole use, since one of their aim is exactly the correction of the everted feet, it can be a phase of adaptation to both kinds of orthoses and a feet re-adaptation to the new position and movements.

1.4.1.3.2. Knee

In frontal plane, knee's movement obtained during first evaluation (Table 11) is different from the expected. It is supposed to observe an adduction movement with the movement degrees increasing until about 60% of gait cycle and following an abduction movement is expected. However graphs, for both knees, show that the degrees increase is not as big as the expected. Also after the first adduction peak, new peaks appear in the graphs, which is not supposed to happen.

Second data collection (Table 27) is better than the first one and closer to the expected in normal gait cycles.

Knee movement on the third data collection (Table 43) is not the expected because, even if for left foot an adduction movement could be identified, it is not as expressive as the expected, and right knee presents an adduction movement at almost 60% of gait cycle instead of abduction movement like on left knee.

Remembering the literature review about the child's disease, it is expected to observe some movement disturbances on knee joint, *genua valgus* (1).

It seems that for barefoot condition the knee movement presents difficulties under frontal plane remain, which is not surprising, since the focus of the orthoses is the ankle and not the knee. Knee's movement adaptations are a consequence of ankle movement modifications (26). And plus the fact that knee's adaptations are a consequence of ankle's changes, between evaluations, modifications on the orthoses have been done, and perhaps it is one of the justifications for the absence of improvements.

1.4.1.3.3. Hip

During the first evaluation (Table 12), hip presents a movement closer to regular.

It is possible to observe on the second data collection (Table 28) a lower range of motion between adduction and abduction without considerable differences between conditions.

Third data collection (Table 44) presents a lower range of motion between adduction and abduction.

In a general way, it does not seem that the hip movement pattern is getting better. Looking to the graphs of each data collection, it seems that for barefoot condition only the movement degrees have decreased. Probably distal segments need more time to show the positive effects (26).

1.4.1.3.4. Pelvis

Pelvis presents along the time a loss in the range of motion just as verified for sagittal plan (Tables 13, 29 and 45).

That can be due to the modifications promoted by the changes on the orthoses that add segments adaptations (26) to the child's gait cycle or correspond to one of the adaptations referred to this kind of population, which is a lower pelvis range of motion in order to walk safely (25).

1.4.1.3. Intersegmental Angles on Transverse Plane

1.4.1.3.1. Ankle

In transverse plane, ankle shows external rotation never turning to internal rotation, as expected in regular gait. During the first data collection (Table 14), curve and peaks were close to the normal gait cycle, it was the best condition observed during this first data collection.

Second data collection (Table 30) presents an almost null movement for right ankle, almost straight until 60% of the gait cycle, instant when a smooth increase in external rotation movement is possible to observe. On the other hand, left ankle presents a proximity to internal rotation with a peak at about 20% of gait cycle and turning to external rotation after 50% of gait cycle.

Third data collection (Table 46) does not present any of the ankles in neutral position, being both in external rotation with changes in the degree of external rotation.

It is possible to observe along the data collection that ankle loses the range of motion and that can be due to the modifications on the orthoses and the beginning of insoles use, that induces new adaptations to ankle on the plantar flexion and dorsi flexion movement but also in the reduction of eversion (26).

1.4.1.3.2. Knee

Knee graphs show, during the first data collection (Table 15), movement in internal rotation when the expected was external rotation, changing at second data collection (Table 31), which shows knee for barefoot condition (almost all the time) in external rotation, which is the expected.

Right knee, during third data collection (Table 47), is always in external rotation, not presenting a considerable range of motion. Left knee is on internal rotation until about 60% of gait cycle, presenting right after that a curve in external rotation. Also the range of motion is smaller than the observed on the first data collection.

This modification can be the result of adaptations promoted by the use of orthoses and insole, which affect directly ankle and as a consequence the knee joint (26).

1.4.1.3.3. Hip

At first data collection (Table 16), barefoot starts the gait cycle at external rotation, being supposed to observe internal rotation right after initial contact that does not happen for barefoot.

Only after 50% of gait cycle it is possible to observe two smooth peaks of barefoot internal rotation for right hip. Left hip for this condition presents internal rotation until about 85% of the gait cycle. Both sides finish the movement in external rotation.

Second data collection (Table 32) presents a decrease of range of motion and the movement starts in external rotation for right hip but not for left hip. It is also possible to observe that, at first data collection, internal rotation was almost never present, and now left hip is always on internal rotation.

Right hip is always on external rotation changing the movement degrees along the gait cycle.

Left hip is always on internal rotation changing the movement degrees along the gait cycle, being possible to observe that after 50% of gait

cycle, barefoot has an external rotation, and in the end of the cycle the internal rotation range of motion decreases almost into neutral position.

At third data collection (Table 48) is possible to observe both hips at external rotation in the initial contact, which was not observed on the second data collection for left hip.

Barefoot is always at external rotation for left and right hip, even when left hip shows two instants in neutral position (when black curve touches the neutral line), maintaining the observed data on the last evaluation for right hip and changing from internal rotation to external rotation for left hip.

1.4.1.3.4. Pelvis

Along the time graphs (Table 17, Table 33 and Table 49) describing pelvis on the transversal plane are in agreement with the expected for a regular gait. Along the time there are no significant modifications.

1.4.2. Orthoses with shoe condition

1.4.2.1. Temporal Distance Results ($\bar{X} \pm \delta$)

		1 st Data Collection	2 nd Data Collection	3 rd Data Collection
Speed (m/s)		1.053	0.870	0.425
Stride Width (m)		0.088±0.026	0.160±0.035	0.213±0.018
Stride Length (m)		1.038±0.046	0.886±0.065	0.585±0.076
Cycle Time (s)		0.986±0.028	1.018±0.214	1.376±0.095
Step Length (m)	Left	0.516±0.031	0.425±0.030	0.284±0.044
	Right	0.532±0.031	0.461±0.057	0.300±0.046
Step Time (s)	Left	0.483±0.009	0.538±0.151	0.671±0.056
	Right	0.493±0.028	0.480±0.064	0.703±0.078
Stance Time (s)	Left	0.560±0.044	0.584±0.096	0.895±0.100
	Right	0.530±0.041	0.634±0.166	0.888±0.097
Swing Time (s)	Left	0.419±0.016	0.438±0.147	0.475±0.075
	Right	0.448±0.037	0.379±0.054	0.474±0.031
Stance Time / Cycle Time (%)	Left	≈ 56.8	≈ 57.4	≈ 65.04
	Right	≈ 53.8	≈ 62.3	≈ 64.53
Swing Time / Cycle Time (%)	Left	≈ 42.5	≈ 43.03	≈ 34.52
	Right	≈ 45.4	≈ 37.2	≈ 34.45
Double Limb Support Time (s)		0.103±0.043	0.198±0.049	0.419±0.132

Table 51 – Temporal Distance Results for Orthoses with Shoe condition

Along the time, it is possible to observe differences in the temporal and spatial parameters.

It is completely clear that **speed** decreases along the time, meaning a more controlled gait, which will influence the other parameters.

Stride width increases significantly between evaluations.

Stride length decreases along the time for about a half when comparing the first and the third evaluations.

Cycle time increases along the time and the same happens with **step time** although **step length** decreases along the time.

Stance time and **swing time** increase along the time with one exception for the right foot in the second evaluation. The relation of time of stance and swing during a gait cycle shows that stance takes more time than swing along the evaluations, according with the expected.

Double support time also increases along evaluations.

Orthoses with shoe condition prove, along the time, to become more mature and safe avoiding the falls stated by the parents in the beginning of this cycle of evaluations.

The only parameter that does not present better results along the time is stride width and this can be due to the adaptations that are being done by the child, not forgetting that orthoses suffered a small modification, increase in the movement degrees in the flexion extension movement, also some modifications in the distal joints can promote this stride width increase.

1.4.2.2. Intersegmental Angles on Sagittal Plane

1.4.2.2.1. Ankle

In the sagittal plane, during first evaluation (Table 3), dorsi and plantar flexion were described by a straight line.

Second data collection (Table 19) shows a different pattern, since it is possible to observe plantar flexion after the contact of foot with the floor and dorsiflexion movement becomes clear for left foot. Also it is possible to observe plantar flexion occurring in the final of stance phase as well as dorsiflexion in the final of gait cycle.

Third data collection (Table 35) presents similar graphs for both feet. It is possible to observe for left ankle, plantar flexion right after the instant when foot contacts with floor. Then both ankles move into a peak of dorsiflexion at about 50% of gait cycle turning to plantar flexion right after, and then it is possible to observe dorsiflexion for right ankle.

The movement under this plane is conditioned by the orthoses and their movement freedom, which means that more movement degree is observed between the 1st and the 2nd data collection as a result of orthoses modifications that were not changed between 2nd and 3rd data collection and at the same time the child started to use insole. The lack of movement should be compensated by knee's flexion during swing phase (25).

1.4.2.2.2. Knee

When observing knee graphs (Table 5), on sagittal plane, it presents an initial extension, turning to flexion like described for regular gait, being those graphs closer to the expected and the same happening during second data collection (Table 21).

Third data collection (Table 37) shows similar pattern for both knees. Starting with a flexion movement followed by extension, in both cases closer to 0°, and then turning into flexion at 40% of the gait cycle, achieving a flexion peak that is followed by extension, almost achieving neutral position.

Orthoses with shoe condition promote an adjusted flexion and extension movement on the knees, even in the degrees of movement, although the first flexion still not going so well.

1.4.2.2.3. Hip

Hip graphs during the first evaluation (Table 7) present a pattern of movement closer to the expected, the same happening during second and third data collections (Table 23 and Table 39).

1.4.2.2.4. Pelvis

Along the time, it is observed that pelvis is positioned backward instead of forward, as stated on literature (26).

During the first evaluation (Table 9), when observing pelvis on the sagittal plane, it is possible to verify that its movement graph is not in agreement to the expected for regular gait, the two peaks and the two valleys were not present.

Along the time the graphs that describe pelvis movement under sagittal plane (Table 25 and Table 41) continue not presenting the two valleys and the two peaks. At the same time losing range of motion, as written before. It can be the result of the adaptations promoted along the time on the lower limb, but can also represent a characteristic of low vision as a way to improve safety to the gait (25).

1.4.2.3. Intersegmental Angles on Frontal Plane

1.4.2.3.1. Ankle

At frontal plane, during first evaluation (Table 10), some movement limitation is possible to observe. It is possible to observe eversion for left ankle in the beginning of the gait cycle but in a general way it is represented by a straight line, which is in agreement with the expectation of movement absence caused by the orthoses' use (26).

During second evaluation (Table 26), it presents an increase of movement degrees if compared with first evaluation, although always close to 0°.

On third data collection (Table 42), right ankle shows an eversion movement right after the contact of foot with the floor returning to inversion. Left foot has a similar pattern although an eversion peak is possible to observe after the 50% of gait cycle returning to inversion right after.

It was expected that the condition orthoses with shoe presented absence of movement (26), but the degrees of movement of this condition increased along the data collections. Some modifications were done to the orthoses, between first and second data collections, increasing the degrees of movement in the sagittal plane.

1.4.2.3.2. Knee

Knee movement under this plane (Table 11, Table 27 and Table 43) is still not in agreement with the expected for a normal gait cycle, although along the time positive improvements were checked, first for left knee (second data collection) and then for right knee (third data collection).

The use of orthoses and insoles like those used by this child (26) had a major effect under the ankle than in the connected joints, meaning that knee will suffer improvements but it could take more time. Also the changes promoted in the orthoses between first and second data collection, that have positive improvements under sagittal plane, could take more time to present it under frontal plane. In addition, the change of orthoses for insole promotes a new adaptation, and positive achievements could need more time.

1.4.2.3.3. Hip

In this plane, during the first evaluation (Table 12), hip presents a movement closer to regular, being possible to observe on the second data collections (Table 28) a lower range of motion between adduction and abduction. Third data collection (Table 44) presents the lowest range of motion between adduction and abduction and it seems that right hip has now a worse performance, which can be the result of insoles use during the last month.

It is known and is stated on the literature review (26), that the use of foot orthoses takes more time to have positive effects under the distal joints.

1.4.2.3.4. Pelvis

Pelvis presents along the time (Table 13, Table 29 and Table 45) a loss in the range of motion just as verified for sagittal plane.

That can be due to the modifications promoted by the changes on the orthoses that add segments adaptations to the child's gait cycle or, according to the chapter visual impairment, correspond to one of the adaptations referred to this kind of population, which is a lower pelvis range of motion in order to walk safely (25).

1.4.2.4. Intersegmental Angles on Transverse Plane

1.4.2.4.1. Ankle

In transverse plane (Table 14 and Table 30), ankle shows a profile which is in agreement with the expected for normal gait cycle, presenting a limitation of movement which is caused by the orthoses

themselves (26). The same happens during the second data collection, being possible to observe a straight line always closer to 0°.

Third data collection (Table 46) presents some degrees of movement in external rotation.

1.4.2.4.2. Knee

Knee's graphs, during first data collection (Table 15), show a movement in internal rotation when the expected was external rotation, which is also possible to observe during second data collection (Table 27).

During second data collection, internal rotation for right knee is clearer than for left knee, which presents a peak at about 60% to 80% of gait cycle.

Right knee during third data collection (Table 47) starts very closer to neutral position, increasing the degrees of movement closer to 50% of gait cycle. After, it moves into external rotation returning to internal rotation right before the end of gait cycle. Left knee presents a more evident internal rotation movement in the beginning of gait cycle, decreasing after 50% of gait cycle and moving into external rotation at approximately 70% of gait cycle, finishing again in internal rotation.

It is possible to observe that, along the data collections, knee loses range of motion, which can be a result of new adaptations since in the beginning internal rotation is observed along almost all the gait cycle, and now it is starting to show a more neutral or external rotation range of motion.

1.4.2.4.3. Hip

In the first data collection (Table 16), the initial contact occurred with hip in external rotation, as expected. The same was observed for second and third data collections (Table 32 and Table 48).

During the first data collection, the internal rotation that should follow it was observed only after 50% of gait cycle for right hip and it only presents two smooth peaks, while left hip presents internal rotation movement after 50% of gait cycle and until 85%.

The gait cycle finishes with orthoses with shoe condition in external rotation, as expected.

During second data collection, right hip is always on external rotation, changing the movement degrees along the gait cycle. It is possible to observe that orthoses with shoe condition, at about 70% of the gait cycle, starts a movement into internal rotation having a smooth internal rotation at about 85% of gait cycle. When compared to the first data collection, it loses range of motion and the time in internal rotation is less.

During second data collection, left hip is always on internal rotation changing the movement degrees along the gait cycle, which was not observed during the first data collection.

During the third data collection, left orthoses with shoe condition curve present internal rotation between 25% and 75% of gait cycle, which is the closest to normal gait cycle observed during this evaluation. Right orthoses with shoe condition is always on external rotation, as observed during the second data collection.

1.4.2.4.4. Pelvis

Along the time the graphs (Table 17, Table 33 and Table 49) that describe orthoses with shoe are similar to those that describe a normal gait cycle. However, compared to the first two data collections, orthoses with shoe lost range of motion along the time and in the last data collection it does not do external rotation.

CHAPTER 4 – CONCLUSIONS

First of all, it is important to notice that the use of orthoses and insole seems to benefit the child's gait pattern, especially at ankle level where both firstly (directly) act, and also at an indirect level on knee, hip and pelvis.

However, it is very difficult to understand the influence of other factors such as therapies and stimulation that are continuous in the child's life, as well as the disease evolution itself. Orthoses of any type should be accompanied by appropriate exercises or therapeutic activities to increase ROM and muscle strength, improve postural alignment, and modify functional activity, as stated by McPoil & Brocato (1985) (30).

It is very important to continue evaluating this child periodically in order to understand what happens as the disease develops, to understand the needs of orthoses adaptations (giving also more time for the child to adapt to them), and to understand how far can this child go.

It is very important to keep this in mind, since there is much work to do and the beneficiaries will be patients with disorders of movement. It is a source of great satisfaction for patients, their parents, and their physicians to know that locomotion and movement disorders are at last receiving the attention they deserve (41).

When we are not able to find studies that permit comparisons, like in the present study case, where it is not possible to compare our data with a similar situation, the study has its own value despite all the limitations (3).

That is why it is also interesting to verify that this is the first case where a positive development is stated, of course due to medicine and therapeutic evolution, knowledge about the disease and how it evolves,

and also due to neural plasticity, individual competence to beat barriers and to adapt to new conditions.

Some problems were found during this study. One of them was the lack of spatio-temporal parameters data and kinematic data for Portuguese population under this age.

The other one is the lack of information about those parameters or the benefits of foot orthoses for disabled population, since the major part of the studies is about normal population (mostly with adults) experiencing abnormal situations.

The marker set is a problem in small children, as described by Sutherland (41), because markers can hit each other, and in this case study the child started to feel bored with that because he felt pain during the period of taking the markers off. Another problem with the marker set is the marker movement that occurs with the skin movements (41), sometimes markers fell, in the last data collection the child did not want the markers so he started to take them off, finding ways to make them fall.

The kinematic methodology has another problem, which is accurate timing of toe-off and heel strike. In that case, the platforms can be helpful, since it will help determining that (41), but in our case, because the child was asked to walk at free velocity, platform information was not achieved.

One of the risks of evaluating small children, according to Whittle (42), is that it is usual to finish without any usable data, and a very upset child.

It might very well be that a real life situation is much more challenging for children with a visual handicap and differences in performance of

gait are revealed. In future studies it would be interesting to provide more challenging real-life situations when looking at gait performance (24).

BIBLIOGRAPHY

1. Toiviainen-Salo S, Linnankivi T, Saarinen A, Mayranpaa MK, Karikoski R, Makitie O. Cerebroretinal Microangiopathy with Calcifications and Cysts: Characterization of the Skeletal Phenotype. *American Journal of Medical Genetics*. 2011: p. 1322-1328.
2. Inman VT. Human Locomotion. *Canad. Med. Ass. J.* 1966 May 14: p. 1047-1054.
3. Sutherland DH, Olshen RA, Biden EN, Wyatt MP. *The Development of Mature Walking* London: Mac Keith Press; 1988.
4. Winter D. *Biomechanics and Motor Control of Human Movement*, Fourth Edition: John Wiley & Sons, Inc.; 2009.
5. Long JT, Wang M, Harris GF. A Model for the Evaluation of Lower Extremity Kinematics with Integrated Multisegmental Foot Motion. *Journal of Experimental and Clinical Medicine*. 2011: p. 239-244.
6. Tolmie JL, Browne BH, McGettrick PM, Stephenson JBP. A Familial Syndrome with Coats Reaction Retinal Angiomas, Hair and nail Defects and Intracranial Calcification. *Eye*. 1988: p. 297-303.
7. Sazgar M, Leonard NJ, Renaud DL, Bhargava R, Sinclair DB. Intravranial Calcification, Retinopathy and Osteopenia: A new syndrome? *Pediatr Neurol*. 2002: p. 324-328.
8. Briggs TA, Abdel-Salam GMH, Balicki M, Baxter P, Bertini E, Bishop N, et al. Cerebroretinal Microangiopathy with Calcifications and Cysts (CRMCC). *American Journal of Medical Genetics Part A*. 2008: p. 182-190.
9. Sheridan MD(aubFM&SA). *From Birth to Five Years, Children's Developmental Progress* Great Britain: Routledge; 1997.
10. Delcomyn F. *Foundations of Neurobiology USA*: W.H. Freeman and Company; 1998.
11. Sutherland DH, Olshen R, Cooper L, Savio LYW. The Development of Mature Gait. *The Journal of Bone and Joint Surgery Vol 62-A N°3*. 1980: p. 336-353.

12. Warraich Z, Kleim JA. Neural Plasticity: The Biological Substrate for Neurorehabilitation. *American Academy of Physical Medicine and Rehabilitation (Suppl. 2)*. 2010: p. S208-S219.
13. Drubach DA, Makley M, Dood ML. Manipulation of Central Nervous System Plasticity: A New Dimension in the Care of Neurologically Impaired Patients. *Mayo Clinic Proceedings*. 2004 June: p. 796-800.
14. Johnston MV. Plasticity in the Developing Brain: Implications for Rehabilitation. *Developmental Disabilities Research Reviews*. 2009: p. 94-101.
15. Gordon AL, di Maggio A. Rehabilitation for Children After Acquired Brain Injury: Current and Emerging Approaches. *Pediatric Neurology*. 2012: p. 339-344.
16. Johnston MV. Clinical disorders of brain plasticity. *Brain & Development*. 2004: p. 73-80.
17. Berencsi A, Ishihara M, Imanaka K. The functional role of central and peripheral vision in the control of posture. *Human Movement Science*. 2005: p. 689-709.
18. Hallemans A, Ortibus E, Meire F, Aerts P. Low vision affects dynamic stability of gait. *Gait & Posture*. 2010: p. 547-551.
19. D'Hont E, Segers V, Deforche B, Shultz SP, Tanghe A, Gentier I, et al. The role of vision in obese and normal-weight children's gait control. *Gait & Posture*. 2011: p. 179-184.
20. Patla AE. Understanding the roles of vision in the control of human locomotion. *Gait & Posture*. 1997: p. 54-69.
21. Hallemans A, Beccu S, Van Loock K, Ortibus E, Truijen S, Aerts P. Visual deprivation leads to gait adaptations that are age- and context-specific: I. Step-time parameters. *Gait & Posture*. 2009a: p. 55-59.
22. Braddick O, Atkinson J. Development of human visual function. *Vision Research*. 2011: p. 1588-1609.
23. Grinter EJ, Maybery MT, Badcock DR. Vision in developmental disorders: Is there a dorsal stream deficit? *Brain Research Bulletin*.

2010: p. 147-160.

24. Hallemans A, Ortibus E, Truijen S, Meire F. Development of independent locomotion in children with severe visual impairment. *Research in Developmental Disabilities* 32. 2011: p. 2069-2074.
25. Hallemans A, Beccu S, Van Loock K, Ortibus E, Truijen S, Aerts P. Visual deprivation leads to gait adaptations that are age- and context-specific: II. Kinematic parameters. *Gait & Posture*. 2009b: p. 307-311.
26. Richards J. *Biomechanics in Clinic and Research*: Elsevier; 2008.
27. Moseley L, Smith R, Hunt A, Gant R. Three-dimensional kinematics of the rearfoot during the stance phase of walking in normal young adult males. *Clinical Biomechanics*. 1996: p. 39-45.
28. Branthwaite HR, Payton CJ, Nachiappan C. The effect of simple insoles on three-dimensional foot motion during normal walking. *Clinical Biomechanics*. 2004; 19: p. 972-977.
29. Nicolopoulos CS, Scott BW, Giannoudis PV. Biomechanical basis of foot orthotic prescription. *Current Orthopaedics*. 2000: p. 464-469.
30. Lockard MA. *Foot Orthoses*. *Physical Therapy*. 1988: p. 1866-1873.
31. Stacoff A, de Quervain IK, Dettwyler M, Wolf P, List R, Ukelo T, et al. Biomechanical effects of foot orthoses during walking. *The Foot*. 2007 September: p. 143-153.
32. Nester CJ, van der Linden ML, Bowker P. Effect of foot orthoses on the kinematics and kinetics of normal walking gait. *Gait & Posture*. 2003: p. 180-187.
33. Stacoff A, Reinschmidt C, Nigg BM, van der Bogert AJ, Lundberg A, Denoth J, et al. Effects of foot orthoses on skeletal motion during running. *Clinical Biomechanics*. 2000: p. 54-64.
34. Shih YF, Chen CY. Effect of insole application on lower extremity kinematics in children with flexible flatfoot. *Gait & Posture*. 2009: p. 107.
35. CLIMEPSI. *Médicos de Portugal*. [Online].; 2013. Available from:

<http://medicosdeportugal.saude.sapo.pt/glossario/oligoamnios>.

36. Milner EC. Motion Analysis Using On-line Systems. In Payton CJ, Bartlett RM. Biomechanical Evaluation of Movement in Sport and Exercise. Oxon: Routledge; 2008.
37. Qualisys. Qualisys Motion Capture Systems. [Online].; 2013. Available from: <http://www.senztech.cc/UploadFiles/20111114111047.pdf>.
38. Stebbins J, Harrington M, Thompson N, Zavatsky A, Theologis T. Repeatability of a model for measuring multi-segment foot kinematics in children. Gait & Posture. 2006: p. 401-410.
39. Bruening DA, Cooney KM, Buczek FL. Analysis of akinetic multi-segment foot model. Part I: Model repeatability and kinematic validity. Gait & Posture. 2012: p. 529-534.
40. Deschamps K, Staes F, Roosen P, Nobels F, Desloovere K, Bruyninckx H, et al. Body of evidence supporting the clinical use of 3D multisegment foot models: A systematic review. Gait & Posture. 2011: p. 338-349.
41. C-Motion. C-Motion Research Biomechanics Wiki-Documentation. [Online].; 2013. Available from: http://www.c-motion.com/v3dwiki/index.php?title=Main_Page.
42. Crenna P, Fedrizzi E, Andreucci E, Frigo C, Bono R. The heel-contact gait pattern of habitual toe walkers. Gait & Posture. 2005: p. 311-317.
43. Sutherland DH. The evolution of clinical gait analysis, Part II Kinematics. Gait & Posture. 2002: p. 159-179.
44. Whittle MW. Clinical gait analysis: a review. Human Movement Science. 1996: p. 369 - 387.

APPENDIX A

Informed Consent

CONSENTIMENTO INFORMADO

Está a ser convidado(a) a participar num estudo de investigação científica que pretende contribuir para caracterizar as disfunções inerentes ao padrão de marcha em crianças com compromisso neuromuscular. Antes de decidir participar, ou não, é importante perceber o porquê desta investigação e os procedimentos que a mesma irá envolver. Por favor leia esta informação cuidadosamente e esclareça todas as dúvidas que achar necessário.

Objectivos do estudo

- (1) Caracterizar do padrão de marcha de uma criança portadora de um síndrome genético complexo e raro "Leucoencefalopatia, calcificações cerebrais e quistos" diagnosticado clinicamente, nas seguintes condições: descalço, com ortóteses e com sapatos.
- (2) Comparar os diferentes padrões mencionados no ponto anterior .

Informação sobre as sessões de teste

Este estudo irá incluir dois tipos de testes:

- (1) Avaliação antropométrica: esta avaliação incluirá massa, altura e altura trocantérica.
- (2) Avaliação da marcha: a marcha será avaliada através de câmaras que captarão a posição de marcadores refletores colocados em proeminências ósseas específicas e nos sapatos e/ou ortótese. Durante esta avaliação serão também utilizadas plataformas de força para medir a força (intensidade e direção) produzida durante o apoio do pé no solo. O participante terá de caminhar à sua velocidade natural. Esta tarefa será repetida algumas vezes em condições diferentes (descalço, com ortóteses e com sapatos)

Para a realização destas sessões o participante deve trazer roupa confortável (sendo aconselhável o uso de calções e t-shirt).

É também recomendado ao participante que se faça acompanhar de todos os exames clínicos complementares de diagnóstico que tenha em sua posse, para que se possa

proceder a um melhor enquadramento da sua condição clínica face à recolha obtida na sessão de teste.

Todos os estudos serão realizados à porta fechada estando apenas presente as pessoas necessárias à recolha de dados, sendo, portanto, garantida a privacidade do participante.

Confidencialidade

A informação obtida neste estudo é confidencial e não será revelada a pessoa alguma sem o seu prévio consentimento, exceto à equipa responsável por este estudo.

Os resultados do estudo utilizados para investigação serão tratados e apresentados de forma inteiramente anónima, e poderão ainda ser reutilizados em estudos futuros.

Participação e Abandono

A participação no estudo é voluntária. É livre de abandonar o estudo em qualquer altura sem qualquer penalidade e podendo ainda, se o desejar, recusar que os dados recolhidos até ao momento sejam publicados.

Danos relacionados com o estudo

Uma vez que as tarefas a desempenhar nas diversas sessões não envolvem atividade física de risco, a probabilidade de sofrer qualquer dano é mínima. No entanto, se tal acontecer, as despesas consideradas razoáveis inerentes a qualquer dano sofrido como resultado direto da sua participação no estudo serão tomadas a cargo pelo Laboratório de Biomecânica da Faculdade de Motricidade Humana.

Termo de responsabilidade

Compreendo perfeitamente todos os procedimentos deste estudo e os riscos inerentes ao mesmo. As minhas dúvidas acerca da participação no estudo foram satisfatoriamente esclarecidas. Caso venha a ter mais alguma dúvida, poderei esclarecê-la junto dos investigadores responsáveis.

Entendo perfeitamente que não sou obrigado a participar no estudo e que posso, em qualquer altura, abandonar o mesmo sem qualquer penalidade.

Fui informado dos meus direitos como participante e sei que, se em alguma altura sentir que os mesmos foram ignorados, negligenciados ou recusados, devo informar o coordenador do Laboratório de Biomecânica da Faculdade de Motricidade Humana (Estrada da Costa, Cruz Quebrada, 1495-688 Cruz Quebrada-Dafundo), que se encarregará de investigar a queixa.

Cruz Quebrada,

Assinatura

Contactos:

Coordenador-investigador do laboratório: Prof. Doutor António Veloso

Investigadores: Lic. Catarina Góis, Lic. Filipa João, Lic. Miguel Martins, Lic. Rodrigo Martins, Lic. Sílvia Cabral, Lic. Vera Bagão, Lic. Vera Moniz-Pereira.

e-mail: labneuromechanics@gmail.com

Tel: 214149127

APPENDIX B

Medical Reports and Parents' Interview

Medical Reports

Child Neurology

Child Neurology
Prof. dr. M.S. van der Knaap
Dr. R.J. Vermeulen
Dr. N. L'Hof
Dr. M.S.M. van Breemen

Your letter dated

Your reference

Date

June 28, 2012

Subject

Deaf-Blind Facetta Ross

Our reference

2012.227

Tel.: +31 20 44 44356

Fax: +31 20 44 40349

Dear Mr. and Mrs. Ross

Thank you for sending the new MRI of your son born 20-02-2008.

A new MRI was obtained on 29-03-2012. The images show signal abnormalities at the level of the pons, middle cerebellar peduncles, thalamus and parieto-occipital white matter, right more than left. There is evidence for calcium deposition in those areas as well. At the level of the thalamus there is a large cyst in the midline. The susceptibility-weighted images show calcium in large areas of the brain, including the basal nuclei, thalamus and border between white and gray matter in the parieto-occipital region, right more than left. There are also calcium depositions in the brain stem and cerebellum.

I must say that as compared to the MRI of 2011 the abnormalities are rather stable. The swelling is less in the right parieto-occipital region. The cyst has not increased in size. Changes in other areas are minimal.

I have not commented on the abnormalities occurring in the right eye. I am no expert on ophthalmic pathology.

All in all I would feel your son is doing well and is rather stable at the moment.

Kind regards,

Marlo S. van der Knaap, MD, PhD
Professor of Child Neurology
VU University Medical Center
P.O. Box 7057
1007 MB Amsterdam
The Netherlands
Phone: +31 20 44 44356
Fax: +31 20 44 40349
E-mail: m.s.vanderknaap@vumc.nl

31st May, 2012

Professor Luis Nunez
Centro Hospitalar de Lisboa Central EPE
Serviço de Genética Médica
Rua José António Serrano
1150-199 Lisboa

Dear Luis

Re: . DOB: 20/02/2008 AG5382

I am writing to confirm that we have identified biallelic CTC1 mutations in your patient. Details are given in the paper by Anderson et al (Nature Genetics AOP 2012 (your patient is annotated as B32)).

I am not sure of the implications of this diagnosis for your patient in longer term. However, I think it would be sensible to be aware of a possible risk of developing gastrointestinal ectasias which, in several patients, have been lethal.

We will continue our studies of Coats plus and would be very pleased to be kept in touch with your patient's progress, and possibly obtain further samples if the family are interested and willing.

Finally, I have to say that these results have been generated on a research basis. For clinical purposes we would recommend confirmation in a diagnostic laboratory.

Thank you very much to you and the family for being so generous with your support of our work.

Yours sincerely



Nick Crow
Professor in Genetic Medicine

PLEASE NOTE: THIS IS A RE-ISSUED LETTER WITH CORRECTED DATE OF BIRTH. THIS LETTER IS TO REPLACE THAT DATED 7th FEBRUARY 2012

RELATÓRIO AVALIAÇÃO CONSULTA DE SUBVISÃO

Doente do sexo masculino de 4 anos de idade, fez avaliação da situação clínica e apresentava os seguintes valores:

VOD – amaurose

VOE $\text{c/c} = 0,32$ (escala dos Es)

Movimentos oculares: Hiperabdução dos músculos pequenos oblíquos

Cover - Endotropia OD com hipertropia

Krimsky - 30Δ BE

Não tem funções binoculares

Visão cromática - identifica todas as cores

Campo Visual OE por confrontação: Campo visual tubular

Visão funcional - Rentabilização muito boa da visão, que lhe permite a identificação de figuras e objetos de vários tamanhos e cores.

Foram analisados vários trabalhos escolares que provam, o que se acaba de descrever, com aproveitamento de toda a folha de base, usada nos trabalhos que desenvolveu, realçando predileção pela cor laranja em todos os desenhos que executa, apesar de, haver comprometimento da coordenação olho/mão devido a problemas motores.

Encaminhamento: Após reunião com a Fisioterapeuta, Professora de Educação Especial, e comigo, Ortopista, concluímos que o menino beneficia de um candeeiro de luz fria que ilumine a área de trabalho (mesa) na escola e em casa. Computador portátil em que lhe serão indicados jogos de estimulação visual, com rato adaptado.

Dra. Maria Emília Mouga
Ortopista



João Paulo Santos
Fisioterapeuta



Ministério da Saúde



**Centro Hospitalar
de Lisboa Central
EPE**

**Hospital Dona
Estefânia**

**Serviço
de
Genética Médica**

Sector Clínico

Especialistas
Prof. Doutor Luis Nunes
(Responsável)
Dra. Teresa Kay
Dra. Marta Amorim
Dr. Rui Gonçalves

R. Jacinta Marto
1169-045 Lisboa

Contactos:
Tel. ++351 21 3126674
Fax: ++351 21 3126869



**Ministério da
Saúde**

Assunto: Relatório médico de

1. Acompanhamento o _____, nascido 20/02/2008, no Serviço de Genética Médica do Hospital Dona Estefânia desde 15/09/2009;
2. Esta criança é portadora da síndrome CRMCC (cerebroretinal microangiopathy with calcifications and cysts), uma doença genética e hereditária, muito rara, identificada em pouco mais de duas dezenas de indivíduos no mundo, sendo o _____ a única criança conhecida em Portugal;
3. A natureza genética da doença foi identificada recentemente, com o contributo de várias famílias, incluindo a família do _____ que fez a doação do material hereditário da criança, do irmão e dos progenitores, às duas equipas que a nível mundial estavam mais avançadas na pesquisa (Reino Unido e Finlândia). Se é verdade que no final também beneficiaram da investigação, merece o meu maior respeito a atitude que sempre tiveram de contribuir com a sua doação para a investigação o que terá vantagem para inúmeros indivíduos e famílias em todo o mundo;
4. A doença manifesta-se essencialmente por quistos cerebrais e calcificações que causam uma perturbação variável no desenvolvimento do sistema nervoso central e lesões dos pequenos vasos (microangiopatia), de início mais evidente na retina, mais tarde noutros órgãos;
5. A doença tem um carácter evolutivo, sendo imprevisível o prognóstico; se inicialmente são afectadas a retina, o que causa graus variáveis de incapacidade, foi descrito que muitas das crianças são afectadas noutros sistemas, como o digestivo, com consequências fatais;
6. A doença do _____ tem manifestações graves, com um atingimento severo do sistema nervoso central e da retina (lesões supra e infratentoriais e calcificações generalizadas no cérebro, que se manifestam por hipertonia central, perturbação do equilíbrio e do desenvolvimento psicomotor) e da retina, que se manifesta por cegueira do olho direito e uma capacidade visual do olho esquerdo estimada em 20% da referência;

7. As anomalias referidas anteriormente traduzem-se por uma incapacidade significativa e desvantagem importante;
8. E se é da maior justiça reconhecer a forma exemplar como os pais e toda a família desta criança se têm empenhado para lhe propiciar o maior conforto e as condições para que tenha acesso aos cuidados de saúde de maior qualidade, não deixa de ser evidente que a criança pode beneficiar muito se o contexto ambiental da sua vida for ajustado ao seu grau de incapacidade;
9. Neste sentido, é da mais elementar evidência reconhecer que é essencial que o ambiente se adapte às necessidades de uma criança diferente mas especial, e não o contrário, que a criança se adapte a contextos que foram conceptualizados para indivíduos sem necessidades especiais;
10. A adequação da habitação é o elemento que se me afigura como mais relevante face ao referido anteriormente, pois é na casa que a criança passa grande parte do seu dia, se integra na vivência familiar, interage e aprende com os pais e restantes familiares; este espaço não é neutro, é valorativo e crucial para a aprendizagem e para a qualidade de vida, independentemente do tempo e da incapacidade que essa vida tenha;
11. Uma criança com as necessidades de saúde especiais carece de uma habitação construída em torno das suas particularidades de saúde, que lhe possa propiciar qualidade de vida e mais bem-estar; esta perspectiva permite-me sugerir, que se tal for possível, _____; possa beneficiar de uma habitação personalizada, construída de raiz, tendo em conta as suas diferentes necessidades;
12. Alguns dos elementos específicos, considerando as necessidades de saúde particulares do _____ que me apraz recomendar em termos de construção de uma habitação apropriada são os seguintes:
 - a) Cobertura extensível desde a porta principal até ao portão principal:
Explicação: melhor acessibilidade e segurança face às condições atmosféricas
 - b) Rampa na entrada da casa:
Explicação: permite uma maior acessibilidade, nomeadamente cadeira de rodas
 - c) Cor dos mosaicos, chão, loiças sanitárias, armários (WC e cozinha) adaptadas à incapacidade da visão:
Explicação: escolher cores que ofereçam contraste entre o piso e o mobiliário / sanitários de forma a criar contraste, facilitando o campo visual
 - d) Colocação de barras de apoio no WC na zona de duche e sanitário:
Explicação: auxiliam a acessibilidade e a movimentação dada a deficiência visual e motora, dando uma maior autonomia
 - e) Espelhos ao longo dos tetos levemente inclinados para baixo:
Explicação: permite que a criança se veja a si próprio ajudando na locomoção
 - f) Colocação de ar-condicionado e aquecedores fixos nos quartos e nas salas:
Explicação: evitar os aparelhos móveis que possuem fio o que dificulta a vida diária de um deficiente motor/visual.
 - h) Instalação de várias tomadas com sistema de segurança:
Explicação: facilita o uso de diversos equipamentos sem fios espalhados pela casa
 - i) Pontos para luz (com opção sonora) de emergência nas áreas de circulação:

Explicação: quando há falta de luz, a pessoa com necessidades especiais circula com segurança evitando situações de perda de controlo e pânico

j) Colocação de interruptores iluminados e/ou com sinalização sonora:

Explicação: são fáceis de localizar, são estimulantes e seguros.

l) Colocação de armários com luzes e/ou sinal sonoro que é accionado quando a porta é aberta:

Explicação: os vários objectos são mais facilmente encontrados pela criança na qualidade de deficiente visual/motor

m) O piso interior e exterior de toda a habitação deverá ser escolhido considerando algumas características importantes: ser antiderrapante, com um design que permita contraste (em termos de cor e de rugosidade):

Explicação: facilita integração sensorial, acessibilidade, maior percepção do espaço / distância (permite que a criança, enquanto deficiente visual, tome consciência da mudança de divisão evitando acidentes)

n) As portas de todas as divisões internas devem ser de correr:

Explicação: propicia uma maior acessibilidade / rentabilidade no espaço

o) Opção por maçanetas do tipo alavanca / com pontas arredondadas e, de preferência, com a fechadura em cima:

Explicação: facilitam a introdução da chave e o manuseio por pessoas portadoras de deficiência motora/visual

p) Colocação de torneiras do tipo mono-comando:

Explicação: evita que a criança se queime e facilita o seu manuseamento tendo em conta que tem tremor não intencional causado pelas lesões cerebrais

q) Colocação de armários de cozinha e WC, mobiliário e portas que façam contraste com a cor das paredes (com quinas arredondadas):

Explicação: proporciona maior acessibilidade dado o contraste e evita lesões

r) A porta exterior, persianas, estores e portões devem ter sistemas de abertura automática:

Explicação: propicia maior acessibilidade / autonomia

s): A iluminação, incluindo a dos candeeiros, deverá ser obrigatoriamente de cor branca e fria:

Explicação: contribui para uma maior rentabilidade dos 20% de visual ainda conservados da criança

t) Vidros interiores e exteriores com tratamento anti-fotossensibilidade

Explicação: contribui para uma maior rentabilidade da visão residual ainda conservada, e preserva o olho dado que a luz causa dor, desconforto e perda de capacidade de focar objectos;

u) As aberturas das portas devem ser mais largas (>80cm), as divisões da casa devem ser amplas, e os rebordos devem ser contrastados com as paredes

Explicação: facilita o acesso à cadeira de rodas ou outros auxiliares de marcha/deslocamento, e previne acidentes;

v) Os armários devem ser baixos, com sistema automático de elevação e recuo

Explicação: permite o acesso e a autonomia;

w) Implementação de cordão de segurança na casa de banho

PROF. ANTÓNIO CASTANHEIRA-DINIS
MÉDICO OFTALMOLOGISTA

PARECER

Foi-me solicitado um parecer sobre as necessidades ambientais que o menino _____ carece devido ao conjunto de incapacidades de que sofre, com realce para as limitações devido à sua deficiência visual.

O menino _____, 5 anos, sofre de doença genética rara afectando o sistema nervoso central e, naturalmente, a retina como expansão periférica do cérebro.

Na observação hoje realizada ao menino Dinis conclui que o menino _____ tem notórias limitações visuais, utilizando unicamente o seu olho esquerdo (o único funcional) com acuidade visual reduzida para 20-30 %, capaz de identificar somente objectos de dimensão considerável e que utiliza predominantemente o seu campo visual temporal esquerdo, adoptando, por isso, posição viciosa da cabeça para melhor se orientar no espaço ambiente e caminhar.

No plano de desenvolvimento sensorial, educativo e integrativo, o menino _____ necessita de apoio especial quer no meio escolar como no meio familiar com ambiente e condições adequadas à sua deficiência sendo mandatório proporcionar-lhe os meios e as condições adequadas para o seu desenvolvimento motor, sensorial, psíquico, cultural e social, representando a visão um sentido fundamental para a percepção do mundo ambiente e, assim, auxiliar o menino Dinis no desenvolvimento global dos aspectos referidos.

Neste sentido, considero que um conjunto de condições ambientais deverão ser proporcionadas ao menino _____ relacionadas com a visão e, naturalmente, com as outras limitações que acompanham a sua doença.

No que se refere à visão, o ambiente de sua casa deve ter condições de iluminação adequadas e específicas, facilitadores para a percepção dos objectos e móveis, devendo-se utilizar diferentes cores para realçar o contraste entre os objectos/mobiliário e realçar o contraste dos móveis sobre as paredes. Deve, ainda, possuir computador, ecrã e teclas ajustáveis à sua capacidade visual bem como outras ajudas técnicas adequadas às suas limitações.

1



Transc: 15848 / 2013080010
Data: **04-04-2013**

Utente:

1

Exames Realizados:

18095R Rm Membros, Cada Segmento Nao Articular

RELATÓRIO

RM Muscular

Protocolo: estudo com protocolo de corpo inteiro no plano coronal em ponderação T1.

Descrição:

Pescoço: sem assimetrias volumétricas musculares, com sinal muscular globalmente homogéneo.

Cintura escapular: deltóide e trapézios simétricos bilateralmente. Coifa dos rotadores sem sinais de infiltração adiposa ou assimetrias volumétricas.

Braço: normal sinal muscular sem sinais de atrofia ou assimetrias volumétricas.

Antebraço: avaliação condicionada dado o protocolo de corpo inteiro.

Tronco e músculos paravertebrais: não se observam assimetrias volumétricas ou do sinal muscular nos vários grupos estudados, sobretudo não se objectivando alterações ao longo do maior eixo longitudinal. Músculos psoas ilíacos sem alterações valorizáveis em termos de volume e sinal.

Cintura pélvica: glúteos simétricos, de sinal homogéneo e planos conservados.

Coxas: compartimento anterior e posterior sem assimetrias volumétricas ou alterações do sinal muscular sugerindo infiltração adiposa focal ou difusa.

Pernas: não se observam assimetrias volumétricas ou de sinal muscular sobretudo a nível do compartimento posterior.

Impressão diagnóstica:

1.O estudo efectuado com protocolo de corpo inteiro e fusão de imagens no plano coronal não revela assimetrias musculares valorizáveis ou sinais de infiltração adiposa significativa.

2.Ressalva-se que a avaliação muscular do antebraço, musculatura das mãos e dos pés não é possível de maneira adequada pelo presente protocolo, necessitando de uma avaliação segmentar dirigida .

Transc: 15848 / 2013080010
Data: **04-04-2013**

Exames Realizados:
18095R Rm Membros, Cada Segmento Nao Articular

Validado por:

Pedro Alves

Pedro Miguel Tojais R Alves
Médico Radiologista

Parents interview

1) By reading the medical reports that you gently give us, it was possible to understand that at the 34 weeks of pregnancy you found that there are some complications with the baby, being referred “intrauterine growth retardation” and later “presence of oligohydramnios”. I would like to know the following:

The baby birth was at?

Between the birth and the 10 months of life, when the psychomotor development retardation was detected, did the doctors focus on the preview information about “intrauterine growth retardation” and later “presence of oligohydramnios”. Do they try to establish some relation between both things?

2) It was described in the medical report that the baby has had “fetal distress”. Can you please describe better what happened.

3) According to the medical report, the first complementary exams done by your child happened when he was 10 months, can you please described what kind of information was given to you.

4) When do you felt that was necessary to find international support?

5) As I understand your child have been submitted to a huge number of surgeries. Can you please briefly describe the dates, the aims and the results achieved?

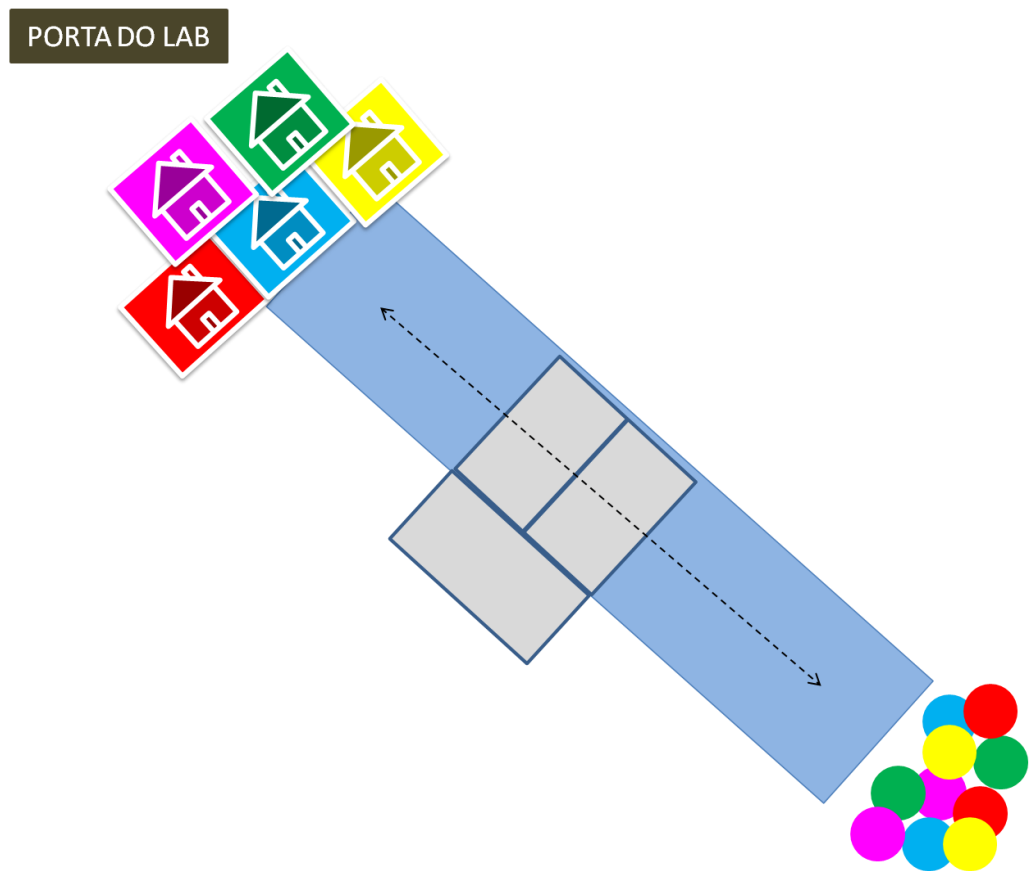
6) Can you please summarize what therapies he has now?

APPENDIX C

Activities

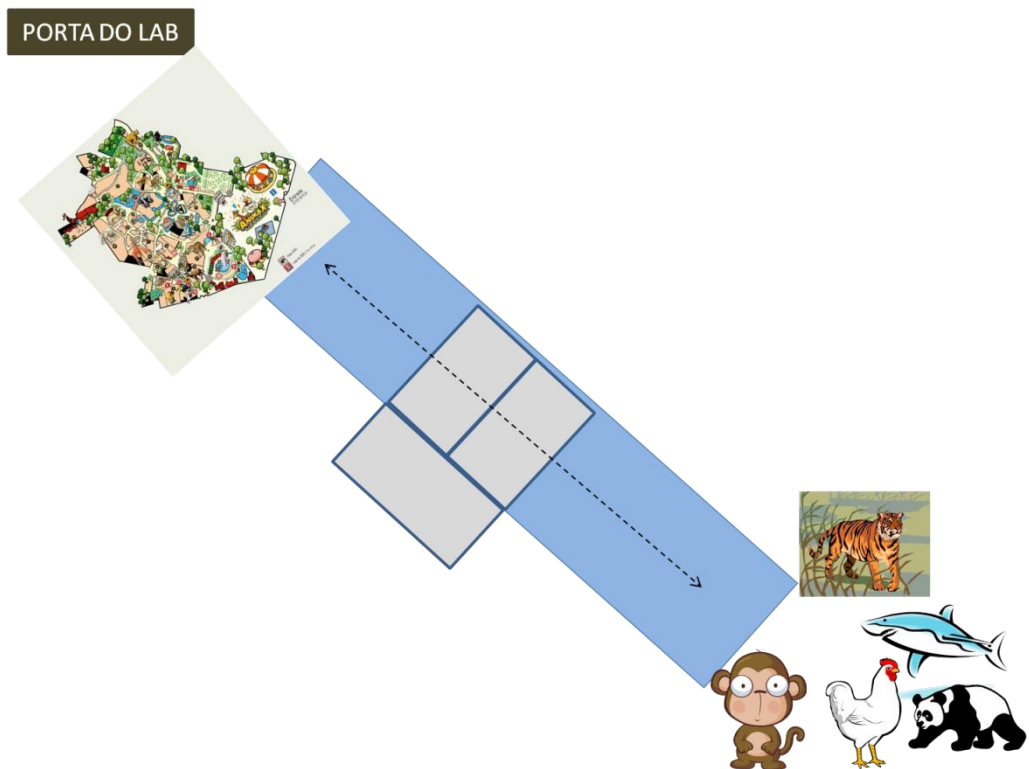
ATIVIDADE 1: CASTELO DE BALÕES

Colocar numa das extremidades do corredor de marcha uma mesa com balões de cores diferentes (amarelos, azuis, verdes, encarnados, roxos, etc) e, na extremidade oposta, colocar os castelos de cartolina com uma janela no centro. O objetivo é a criança pegar num dos balões, atravessar o corredor a caminhar e no outro lado fazer passar o balão pela janela do castelo que tem a mesma cor. Repetir o procedimento até terminarem todos os balões.



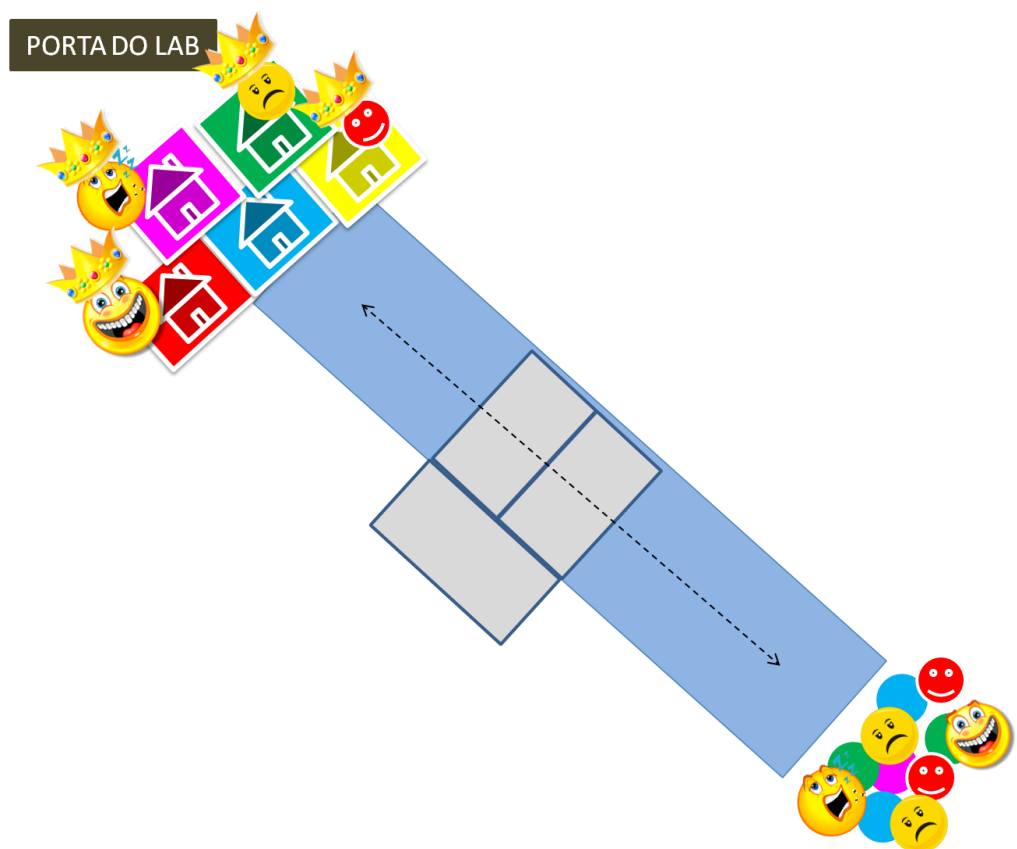
ATIVIDADE 2: OS ANIMAIS FUGIRAM DO ZOO!

Colocar numa das extremidades do corredor de marcha uma mesa com vários animais (podem ser recortes de imagens que se colam em cartão) e, na extremidade oposta, colocar um tabuleiro onde estão desenhados os “habitats” de cada um no zoo mas sem os animais (pode deixar-se apenas o espaço com a forma do animal que falta). O objetivo é a criança pegar num dos animais, atravessar o corredor a caminhar e no outro lado colocar o animal no respetivo “habitat”. Repetir o procedimento até terminarem todos os animais.



ATIVIDADE 3: O REI FELIZ OU DORMINHOCO?

Desenhar diferentes expressões faciais em quadrados de cartolina que possam ser acrescentados ao castelo da atividade 1: passam a ser “o castelo do Rei Feliz”, ou “o castelo do Rei Dorminhoco”. Aproveitar os mesmos balões e desenhar nestes as mesmas expressões. Desta vez, o objetivo é a criança pegar num dos balões, atravessar o corredor a caminhar e no outro lado fazer passar o balão pela janela do castelo do Rei que tem a mesma expressão (as cores não contam!). Repetir o procedimento até terminarem todos os balões.

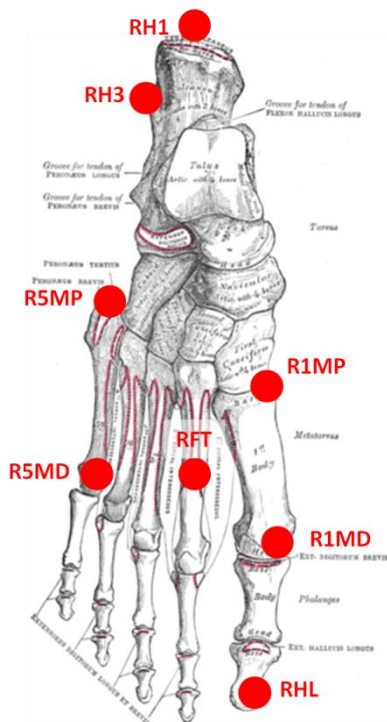
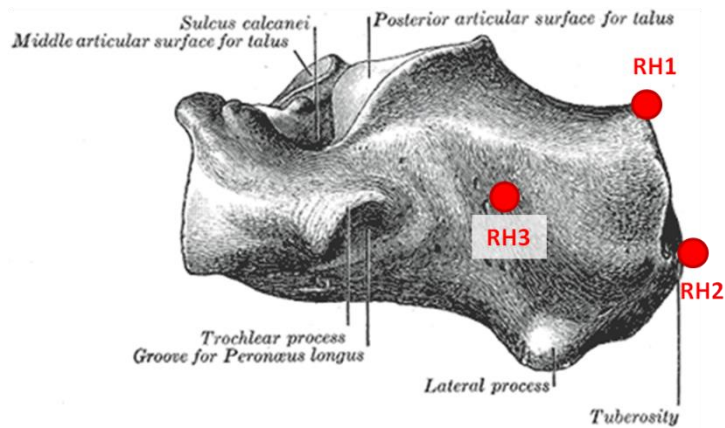


APPENDIX D

Marker placement

Barefoot

Right	Left	Notes
LAC	RAC	Follow the scapula's spine towards the shoulder joint, until you find the acromial angle. Follow the clavicle until you find the acromio-clavicular joint line (small depression). The marker should be placed on the midpoint of this line, which is obliquely oriented (shoulder top).
C7		C7 is the most prominent vertebra in the cervical region (when you see two prominences, C7 is the one that does not disappear with head flexion).
STRN1 - 2		Vertically aligned on top of the sternum (for tracking only)
RASI S	LASI S	Palpate along iliac crest in the anterior direction until you find the anterior superior iliac spine (flat surface after the end of the crest).
RPSI S	LPSI S	Palpate along iliac crest in the posterior direction until you find the posterior superior iliac spine (prominence at the posterior end of the crest).
RTH 1-4	LTH 1-4	Thigh cluster placed according with wobbling mass and visibility.
RLK	LLK	Placed on the lateral epicondyle of the knee - find the mid distance of the ROM, as the epicondyle will change position during the motion.
RMK	LMK	Placed on the medial epicondyle of the knee - find the mid distance of the ROM, as the epicondyle will change position during the motion.
RSK 1-4	LSK 1-4	Shank cluster placed according with wobbling mass and visibility
RLA	LLA	Placed on the lateral malleolus along an imaginary line that passes through the transmalleolar axis.
RMA	LMA	Placed on the medial malleolus along an imaginary line that passes through the transmalleolar axis.
RH1	LH1	Placed on the posterior calcaneus just after the "free" part of the Achilles tendon
RH2	LH2	Placed on the calcaneus tuberosity. Together with the previous marker, these markers will define the alignment of the calcaneus.
RH3	LH3	Marker just for tracking: Placed on the lateral side of the calcaneus between the line of the posterior markers and the line of the lateral malleolus markers
RFT	LFT	Marker just for tracking and to define calcaneus' sagittal plane: On top of the second metatarsal, at the same height of RH1
R1M P R5M P	L1M P L5M P	Place on top of the proximal 1st and 5th metatarsal head, Just after (distally) the tarsometatarsal joint
R1M D R5M D	L1M D L5M D	Place on top of the distal 1st and 5th metatarsal head, Just before (proximally) the joint between the metatarsal bone (1 st and 5 th) and the phalanges.
RHL	LHL	Placed on top of the hallux



Orthoshoe and InsoleShoe

ORTHOSHOE MARKER SETUP		
Right	Left	Notes
LAC	RAC	Follow the scapula's spine towards the shoulder joint, until you find the acromial angle. Follow the clavicle until you find the acromio-clavicular joint line (small depression). The marker should be placed on the midpoint of this line, which is obliquely oriented (shoulder top).
C7		C7 is the most prominent vertebra in the cervical region (when you see two prominences, C7 is the one that does not disappear with head flexion).
STRN1 - 2		Vertically aligned on top of the sternum (for tracking only)
RASIS	LASIS	Palpate along iliac crest in the anterior direction until you find the anterior superior iliac spine (flat surface after the end of the crest).

RPSIS	LPSIS	Palpate along iliac crest in the posterior direction until you find the posterior superior iliac spine (prominence at the posterior end of the crest).
RTH 1-4	LTH 1-4	Thigh cluster placed according with wobbling mass and visibility.
RLK	LLK	Placed on the lateral epicondyle of the knee - find the mid distance of the ROM, as the epicondyle will change position during the motion.
RMK	LMK	Placed on the medial epicondyle of the knee - find the mid distance of the ROM, as the epicondyle will change position during the motion.
RSK 1	LSK 1	Shank marker placed anteriorly and below the tibial tuberosity
RSK 2	LSK 2	Shank marker placed laterally and above the malleolus
RSK 3	LSK 3	Shank marker placed posteriorly, above the malleolus and slightly below SK2 marker
OrthoR LA	OrthoL LA	Placed on the lateral malleolus along an imaginary line that passes through the transmalleolar axis.
OrthoR MA	OrthoL MA	Placed on the medial malleolus along an imaginary line that passes through the transmalleolar axis.
OrthoR H1	OrthoL H1	Placed on the posterior calcaneus just after the "free" part of the Achilles tendon
OrthoR H2	OrthoL H2	Placed on the calcaneus tuberosity. Together with the previous marker, these markers will define the alignment of the calcaneus.
OrthoR H3	OrthoL H3	Marker just for tracking: Placed on the lateral side of the calcaneus between the line of the posterior markers and the line of the lateral malleolus markers
OrthoR FT	OrthoL FT	Marker just for tracking and to define calcaneus' sagittal plane: On top of the second metatarsal, at the same height of RH1
OrthoR 1MP OrthoR 5MP	OrthoL 1MP OrthoL 5MP	Place on top of the proximal 1st and 5th metatarsal head, Just after (distally) the tarsometatarsal joint
OrthoR 1MD OrthoR 5MD	OrthoL 1MD OrthoL 5MD	Place on top of the distal 1st and 5th metatarsal head, Just before (proximally) the joint between the metatarsal bone (1 st and 5 th) and the phalanges.
OrthoR HL	OrthoL HL	Placed on top of the hallux