UNIVERSITAT POLITÈCNICA DE CATALUNYA
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Facultat d'Òptica i Optometria de Terrassa

GRAU EN ÒPTICA I OPTOMETRIA

TREBALL FINAL DE GRAU

VISUAL ASPECTS OF THE POPULATION WITH DOWN'S SYNDROME

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21 D'OCTUBRE DEL 2015

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I per a què consti, signo/em aquest certificat.

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GRAU EN ÒPTICA I OPTOMETRIA

VISUAL ASPECTS OF THE POPULATION WITH DOWN'S SYNDROME

INTRODUCTION:

Down syndrome (DS) is a chromosomal alteration with a third copy of chromosome 21, associated with mental disabilities and physical anomalies, which affect the eyes and visual function, as well as other parts of the body.

OBJECTIVES:

To produce an extensive literature review of the main vision anomalies found in DS individuals and to analyse and compare the data obtained from a group of DS subjects examined.

METHODOLOGY:

A routine visual examination was designed and applied to a group of 22 subjects with DS from Fundació Down Lleida. Visual acuity was measured with Visual Acuity Chart Test Light House at 3m. Frisby stereoacuity test was used to assess the state of binocular vision. Over refraction was used to ensure good refractive correction. Accuracy of accommodation response was assessed by determining the lag of accommodation with the Nott technique and a modified Nott at 20 cm. Colour vision was also tested with Ishihara (38 plates chart).

RESULTS:

The results show lower binocular visual acuity (0.5±0.17 decimal scale), higher incidence for refractive error, being hyperopia the most common refractive error in females, and a greater lag of accommodation in DS subjects compared to normal. With-the-rule and oblique astigmatism are the most frequent. Stereopsis is poor and no colour vision deficiencies were found in the sample studied.

CONCLUSIONS:

The results observed are in general consistent with those found in the literature. DS subjects show an increased incidence of refractive errors and poor accommodative response, which may interfere with their learning process. It is therefore, of utmost importance that these subjects are examined as early as possible in order to detect and correct any visual deficiencies.

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GRAU EN ÒPTICA I OPTOMETRIA

VISUAL ASPECTS OF THE POPULATION WITH DOWN'S SYNDROME

INTRODUCCIÓ:

El Síndrome de Down (SD) és una alteració cromosòmica amb una tercera còpia del cromosoma 21 associada amb discapacitat mental i anomalies físiques, que afecten els ulls i la funció visual, així com altres parts del cos.

OBJECTIUS:

Realitzar una extensa recopilació bibliogràfica de les principals anomalies de la visió que es troben en individus SD, i analitzar i comparar les dades obtingudes a partir d'un grup de subjectes amb síndrome de Down examinada.

METODOLOGIA:

Un examen de rutina visual va ser dissenyat i realitzat a 22 subjectes amb SD de la Fundació de Down Lleida. L'agudesa visual es va mesurar amb el test d'agudesa visual Light House, a 3m. El test d'estereoagudesa de Frisby es va utilitzar per avaluar l'estat de la visió binocular. Es va dur a terme la sobreerrefracció dels subjectes a fi d'assegurar una bona correcció refractiva. La resposta acomodativa es va avaluar mitjançant la determinació del desfasament d'acomodació amb la tècnica de Nott i Nott modificat a 20 cm. La visió del color també va ser examinada amb el test d'Ishihara (38 cartes).

RESULTATS:

Els resultats mostren una agudesa visual baixa binocular (0.50±0.17 escala decimal), major incidència de defectes de refracció, sent la hipermetropia l'error refractiu més comú en les dones, i un major retard d'acomodació en subjectes amb SD en comparació a normals. Astigmatismes a favor de regla i oblics són els més freqüents. L'Estereopsis és pobre i no es van trobar deficiències de la visió del color a la mostra.

CONCLUSIONS:

Els resultats observats són en general consistents amb els trobats en la literatura. Els individus amb SD mostren una major incidència de defectes de refracció i pobre resposta acomodativa que poden interferir amb el seu procés d'aprenentatge. És per tant, molt important que aquests s'examinin tan aviat com sigui possible, per tal de detectar i corregir les possibles deficiències visuals.

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GRADO EN OPTICA I OPTOMETRIA

VISUAL ASPECTS OF THE POPULATION WITH DOWN'S SYNDROME

INTRODUCCIÓN:

El Síndrome de Down (SD) es una alteración del cromosoma 21 asociado a discapacidades mentales y anomalías físicas que afectan a los ojos y a la función visual, como en otras partes del organismo.

OBJETIVOS:

Realizar una extensa recopilación bibliográfica de las principales anomalías de la visión que se encuentran en individuos SD, y analizar y comparar los datos obtenidos a partir de un grupo de sujetos de SD examinados.

METODOLOGÍA:

Un examen visual de rutina fue diseñado y realizado a un grupo de 22 sujetos con síndrome de Down de la Fundación de Down Lleida. La agudeza visual se midió con el test de agudeza visual Light House a 3m. El test de estereoagudeza de Frisby se utilizó para evaluar el estado de la visión binocular. La sobrerrefracción se utilizó para asegurar una buena corrección refractiva. La precisión de la respuesta acomodativa se evaluó mediante la determinación del desase de acomodación con la técnica de Nott y Nott modificado a 20 cm. La visión del color también fue examinado con el test de Ishihara (38 cartas).

RESULTADOS:

Los resultados muestran una agudeza visual binocular baja (0.50±0.17 en escala decimal), una mayor incidencia de defectos de refracción, siendo la hipermetropía el defecto refractivo más común en mujeres, y retardo de acomodación, especialmente en distancias más cercanas, en sujetos con Síndrome de Down en comparación a sujetos normales. El astigmatismo a favor de regla y oblicuo son mas incidentes. La estereopsis es pobre y no se observaron deficiencias de la visión del color.

CONCLUSIONES:

Los resultados observados son en general consistentes con los encontrados en la literatura. Los sujetos con SD muestran una mayor incidencia de defectos de refracción y pobre respuesta acomodativa que pueden interferir con su proceso de aprendizaje. Es por lo tanto, de suma importancia que estos individuos se examinan tan pronto como sea posible con el fin de detectar y corregir las posibles deficiencias visuales.

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EXTENSIVE SUMMARY

VISUAL ASPECTS OF THE POPULATION WITH DOWN'S SYNDROME

INTRODUCTION

Down syndrome (DS) is a genetic disorder characterized by the presence of a third copy of chromosome 21, also known as trisomy 21. It is one of the most common chromosomal abnormalities in new-borns, with an incidence of 10 for every 10,000 births in the world (M.E. Weijerman et al.; 2010).

This population presents physical anomalies and mental disabilities, being vision one of those affected.

This dissertation consists of a literature review compilation of the studies conducted and published on the visual problems of this population, in order to get in depth knowledge of the visual characteristics that are more frequently present in this population.

At the end of the dissertation, there are the results I was able to realize thanks to the experience gained during my stay at the School of Optometry and Vision Science, which I was able to conduct a clinical trial in subjects who have DS. This was possible with the collaboration of the Fundació Down Lleida, which allowed me to put into practice my experience by performing optometric exams to a group of 22 Down syndrome subjects.

The analysis and classification of the reviewed literature allowed us to know:

- Which visual aspects are affected in Down syndrome population and what are the main differences with individuals who do not have the syndrome.
- What visual results or responses can be expected when examining the visual system in these subjects.
- Which are the most suitable tests and assessment methods to perform a visual examination in these individuals.

OBJECTIVES

The main goals for this study are:

- Literature review:

To produce an evaluative report of the most common vision deficiencies or anomalies found in DS individuals by describing, summarizing, evaluating and clarifying the literature.

- Experience abroad:

To take advantage of the experience learn at the short residency spent at the specialty clinics of the School of Optometry and Vision Science.

- Clinical study:

To perform a visual exam and collect data from a group of Down syndrome subjects (N=22) in order to compare our results to those in the literature. Also, to put in practice the skills and abilities learnt during my short residency at the Down syndrome specialty Clinic directed by professor M. Woodhouse, a world well known expert in this area.

- Improvement of English language:

To improve my English communication skills by writing and presenting this dissertation in English.

VISUAL FUNCTION

The main areas of visual function studied where visual abnormalities are present in Down syndrome individuals are:

- Visual acuity and contrast sensitivity:

Studies such as Little et al; (2007), J.M. Woodhouse et al; (1996) among others, have shown that visual acuity (VA) in young children who have Down syndrome is similar to that of young individuals who do not have syndrome when they are very young (up to 3-4 years of age). As children get older, VA improves to a maximum in normal children, whereas DS children do not improve as much leaving them with a lower VA compared to normals. These studies also indicate that the Visual acuity value average that these individuals are able to achieve is 0.4-0.5 monocularly and 0.6 binocularly, regardless of the refractive error and sex.

Regarding to the contrast sensitivity function (CSF), this is below the normal limits compared to normal population and this reduction is more acute at higher frequencies.

- Refractive error:

Some studies (Al-Bagdady M. et al. 2010, Watt et al.; 2014) have shown that there is a notable difference in the incidence of refractive errors between the two populations (DS and normal population). There exist differences in refractive error since early childhood, which increases with age. A possible hypothesis for that is the lack of emmetropization in subjects with Down syndrome (J. M. Woodhouse; 2005, T. Watt et al.; 2014, M. Al-Bagdady et al.; 2010).
In our study, hyperopia has shown to be more prevalent among individuals with Down syndrome and more in females than males.

Astigmatism also follows a similar pattern to the spherical refractive error, as it does not diminish with age. Normally, astigmatism follows a pattern of variation over the years, with wit-the-rule astigmatism from birth, with tendency towards oblique astigmatism variation with age (M. Al-Bagdady et al.; 2010, Woodhouse et al.; 2000). Slanted palpebral fissure of the eyelid has been suggested as a predictive cause for oblique astigmatism.

Anisometropia is also more common in individuals with Down syndrome.

- Accommodative function:

Seventy percent of Down syndrome population have low capacity to accommodate, showing lag of accommodation at close distances. This accommodation is insufficient even from 3 months of age, (J.M. Woodhouse et al.; 2005).

The causes for the reduced accommodative function are not really known, but there exist several hypothesis. The thickness of the lens, which affects the total power of the lens, maybe the reason for a reduced capacity to accommodate (HØvding and Haugen; 2001). Another reason suggested would be due to a general malformation of the parasympathetic nervous system (J. M. Woodhouse; 2005). However, none of these hypotheses and others are sufficiently strong nor conclusive and need more research in this field.

- Disorders of binocular vision:

Strabismus and nystagmus are binocular vision disorders commonly found in people with Down syndrome.

Refractive error or poor accommodation are discarded as the only possible causes for strabismus, probably there are others, not yet know.

Endodesviations are the most common type.

The incidence of nystagmus is also high. One of the causes of nystagmus incidence is believed to be due to heart defects (M. E. Weijerman et al.; 2010), as it is known that they also have other systemic conditions.

- Ocular health:

Brushfield spots, blepharitis, cataracts and keratoconus are the most common ocular disorders in people with Down syndrome (T. Watt et al.; 2014, M. E. Weijerman et al.; 2010, Creavin A et al.; 2009, B Haargaad et al; 2006)

Congenital cataracts and acquired cataracts are the most common type in these individuals. (M. E. Weijerman et al.; 2010, A.J. Adams et al.; 1993, B Haargaad et al; 2006)

Keratoconus is more common in adult ages and it is rare to find cases of children with keratoconus (M. E. Weijerman et al.; 2010, J.A. Little et al.; 2007).

- Colour vision:

As we have seen in previous sections, people with Down syndrome have more affinity for eye and vision abnormalities. So, the same happens to colour vision, with a slightly higher incidence in Down syndrome subjects than in normal people (A.J. Adams et al.; 1993, Pérez-Carpinell J. 1994). The default colour vision is common Protanopia (A.J. Adams et al.; 1993).

		Normal population incidence	DS population incidence
Strabismus		2-4%	20- 47%
Nystagmus		0.001%	11-29%
Cataracts	**Acquired**	1%	3 – 15%
	Congenital	1-5%	4 – 7%
Keratoconus		0.0025%	9-18%
Colour vision deficiency		1.5%	18%

Table 1.1: Comparative table for the incidence of common ocular and visual anomalies between normal and DS subjects.

CLINICAL ASSESSMENT AND METHODOLOGY

Due to the high incidence of eye problems in Down syndrome from birth, routine eye exams are recommended annually.

The most common examinations for a visual routine examination in Down's syndrome is a good anamnesis, visual acuity, refractive error correction, examination of the role and function of binocularity and accommodation, assessment of stereopsis and eye health. Examination of colour vision is also used at certain times.

Visual acuity can be measured with several tests. In our study we used Visual Acuity Chart Test Light House located at 3m. The subjects were asked to identify the figures in the chart while wearing the appropriate correction. In case of non-verbal communicative subjects, these were given a matching card (including the same optotypes) and were asked to match the symbol to the corresponding symbol of the chart.

The state of binocular vision is a field that can be examined quickly and easily with tests like the cover test (CT), ocular motility (MOT) and the near-point convergence (PPC).

In our study, in order to evaluate the accommodative response all subjects where checked to ensure a good refractive correction. Although, the refractive error may be evaluated with the Mohindra technique or under cyclopaedia, in this project this was not realized, instead over refraction was performed to save time in screening. This is carried out by placing a + 2D lens in the trial frame and observing the reflex from the retinoscope. Once refractive error was corrected, the accommodative function was evaluated.

The technique used to evaluate the accommodative response to a fixating target was dynamic retinoscopy (Nott). In this project, the accommodative response was evaluated not only at 40cm (normal distance for Nott), but also but also at 20cm. The fixating target had small accommodative details that attrated attention and ensured stimulating the accommodative response. Afterwards, neutral point was achieved by moving backwards when with movement was found. If against movement was found this was simply recorded. Results were noted in all cases.

The fundus and ocular surface to rule out cataracts and other eye disorders were also observed. This is of high importance to perform eye health examination.

Tests such as colour vision and stereoacuity can provide additional information on these subjects' vision. In this project, we used the Ishihara test (38 plates) for colour vision and the Frisby stereotest, which is an appropriate and simple test for children and mentally handicapped.

Prior to the visual assessment all participants had the consent forms signed by their parents or tutors.

RESULTS

Once obtained and analysed the data of the clinical (N=22) with Down syndrome, aged from 6 to 40 years old, from Foundation Down Lleida, we found that mostly all results obtained at the clinical assessment are in line with the results found in literature for Down's syndrome population.

For visual acuity, mean values of $0.29(\pm 0.10)$ – $0.29(\pm 0.12)$ monocularly and $0.50(\pm 0.17)$ binocularly were achieved, and the refractive error did not show any influence for better or poorer visual acuity.

Regarding the average refractive error, spherical equivalent refraction (SER) was obtained on all DS subjects. The spread of refractive error for hyperopia was about $+2.39 \pm 1.65$D and $+2.879 \pm 1.52$D for the right and left eye, while the mean myopic refractive error (SER) is -3 ± 1.85D and -2.709 ± 1.69D respectively.
Only few subjects presented high refractive error (SER) greater than +5D or -6D and all cases were all females. We have also seen in this study that women tend to be more hyperopic than men, and men more myopic.

As for astigmatism, it was present in almost all subjects. With-the-rule astigmatism and oblique astigmatism were the most common type of astigmatism in this population. Considering astigmatisms ≤ 0.75DC irrelevant, astigmatisms of 1-2D are the most frequent. This study found 4/14 subjects with DS who had significant oblique astigmatism (45º and 135º degrees).

Our results show that lag of accommodation was greater for Nott modified exam (20cm distance) than for Nott (40cm). This is in line with other studies (Rouse et al.; 1984, Woodhouse et al.; 1993, M. Cregg et al.; 2001) where accommodation has shown to be affected in almost all subjects and where lag of accommodation is greater at closest distances.

No subjects with Down syndrome in our study were found to present colour vision anomaly. Ishihara 38 plates colour vision used in this examination. Meanwhile, Frisby stereotest has shown that the stereo acuities are lower than what was expected at the beginning.

Accommodative strabismus, nystagmus, blepharitis, cataracts and brushfield spots have been found in our sample of Down syndrome subjects from Fundació Down de Lleida, being accommodative strabismus the most frequent ocular condition found (12%), followed by cataracts, blepharitis and nystagmus with an incidence of (8%) and a 4% for brushifield spots.

DISCUSSION AND CONCLUSION

In general, the results observed in our study are consistent with the results in the literature.

Our study obtained similar mean values for visual acuity as studies such as Little et al; (2007), Ahmad et al; (1976) among others. The type and grade of the refractive error or sex did not influence in the visual acuity.

Regarding contrast sensitivity function (CSF), in Down syndrome subjects it is usually lower than the normal subjects, following the same curve, but at lower limits. However, in this study contrast sensitivity was not evaluated due to time constants.

There exist differences in the incidence of refractive errors between subjects with Down syndrome and normal individuals, and this tends to be more remarkable over the years. Studies as T. Watt et al.; (2014), Courage et al.; (1997), J.M. Woodhouse et al.; (1993), Al-Bagdady; (2011), Ljubic A et al; (2011), agree with a failure in the process of emmetropization in Down syndrome, which also affects to the incidence of astigmatism.

Hyperopia refractive error is more common in Down syndrome (J.M. Woodhouse et al. 1997) and is greater in women than in males (Neisha M. Rodriguez et al.; 2013, Daniel Monsálvez-Romín.; 2015). This is in line with the results of our study, which could prove that there is a higher incidence of hyperopia than myopia, with more involvement in women than in men.
Astigmatism is also characteristic in Down syndrome subjects, and with-the-rule astigmatism and oblique astigmatism are the most common type of astigmatism (Al-Bagdady; 2010) as it has been shown in our study with 1/3 of frequency for with-the-rule astigmatism and 1/3 for oblique astigmatism. The other 1/3 did not present significant astigmatism.

Large majority for lag of accommodation was observed in our sample of Down syndrome, and the incidence for it was greater at closest distances, as could be seen at Nott modified distance (20cm). The same conclusion was extracted from studies like Cregg et al. (2001), H. Olav Haugen et al (2001), where the same statements for lag of accommodation were found.

Finally, 20% subjects from the sample presented blepharitis, nystagmus, cataracts or brushfield spots, which it has been demonstrated to be greater in people with DS than in normal subjects and 12% strabismus (B. Haargaad et al.; 2006, Creavin A et al.; 2009, T. Watt et al.; 2014).

Despite the small sample of DS subjects from Fundació Down Lleida at my personal clinical experience, results are line with literature collected previously. However, further research is needed in this area because it is a very extensive and with still much to learn yet. It is therefore, of utmost importance that these subjects are examined as early as possible in order to detect and correct any visual deficiencies.

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1 INTRODUCTION

Down's syndrome (DS) is one of the most common chromosomal abnormalities in new-borns affecting 10 per 10,000 live births throughout the world. These subjects tend to suffer from several pathologies, physical alterations and deficiencies in which vision is one of the parts affected.

Down syndrome population suffer from high prevalence of hyperopia, myopia and astigmatism compared to normal subjects. Dysfunctions with binocularity are also common, not to mention that there is a higher prevalence for ocular features such as cataracts, blepharitis and more.

All the information published about visual findings in Down syndrome will be summarized in this project, focusing on what visual acuity they are able to reach and how is their contrast sensibility affected, the variability of refractive error they have and how it develops and a brief explanation for management of these patients. This dissertation will also focus on accommodation, which is one of the most discussed aspects of vision deficiencies in Down's syndrome and finally the state of binocular vision and ocular health will be discussed, with some examples of the most common conditions in Down syndrome population.

This project focuses on a compilation of various studies carried out by professionals of recognized prestige in this field, such as J.M. Woodhouse from Cardiff University School of Optometry.

It is the aim of this study to suggest the best clinical methods for correct visual assessment in Down syndrome subjects, as well as to explain how to conduct the examination, which I had the opportunity to learn and practise during an exchange at Cardiff University School of Optometry where J.M. Woodhouse and her research team have specialty Clinic.
The results obtained from the clinical assessment of a small sample of Down syndrome subjects have been collected and compared to the result from the literature.

2 OBJECTIVES

Different studies have shown that persons affected by Down syndrome have some visual problems associated. These problems can be systemic or physical. This study is a literature review of the main visual problems, which affect Down syndrome population, as well as, a clinical investigation on the refractive errors and accommodative response found in a population of Down syndrome subjects.

The main goals for this project are divided in:

- Literature review:

To collect information of the chromosomal alterations of Down syndrome related to the vision deficiencies. With this, to describe the ocular, physical characteristics and genetic of Down syndrome population and to know the prevalence of the visual problems compared to normal population.

- Experience abroad:

Take advantage of the possibility to study at the Cardiff University School of Optometry and be able to participate with professionals in Down syndrome, such as J. Margaret Woodhouse, during my stance there and get the necessary knowledge and skills to be able to examine adequately a person with Down syndrome.

- Clinical study:

To collect data from a visual exam to a small group of DS subjects (N=22) in order to compare results from visual acuity, refraction, binocular vision, accommodation response, colour vision and stereopsis with those from other studies after the analysis and discussion of results.
Also, to put in practise the skills and abilities learned during my short residency at Down syndrome specialty Clinic directed by professor Woodhouse, a world well known expert on this area at Cardiff University School of Optometry

- Improvement of English language:

Finally, in order to improve my proficiency in English, I have decided, together with my tutors, to write and expose this dissertation in English.

3 VISUAL DEVELOPMENT

The development of the human eye is a complex process of an orderly succession of different events.

This chapter will discuss the development of the organ of the vision from the point of embryogenesis, the development of the visual acuity and the refractive variation that can be found throughout infancy age in normal subjects. At the end, a brief explanation of the oculomotor system will be explained too.

3.1 EMBRYOGENESIS

Embryogenesis starts the 22nd day of gestation, with an embryo of a length of 2mm. The process of gestation takes about 38 weeks, with an embryonary process of four weeks in which the different types of primary tissues develop. It is a delicate process in which any problem can lead to the loss of the embryo. Afterwards, during the next 5 to 8 weeks, in the embryonary process the majority of organs develop, including the basic structures of the eye (ecto, meso and endoderm tissues). This is when ocular anomalies can occur, such as anoftalmia, coloboma, ... After this; in the fetal period (8 to 39 gestation weeks) internal ocular tissues grow and develop. In this period of time, ocular anomalies will occur in specific tissues.

Optic vesicles emerge on the 5th week from the formation of neural ectoderm, starting to invaginate inducing the formation of the lens placode. Blood vessels from the mesoderm will form the choroids, the sclera and extraocular muscles.
The principal anomalies caused during the ocular development are: malformation of the neural tube and optical vesicle causing anoftalmo and microftalmo, coloboma, aniridia, congenital cataracts, goniodisgenesis, congenital glaucoma, retinopathy of prematurity (ROP), congenital nystagmus, ...

3.2 VISUAL ACUITY

New-born babies have very poor central vision and they gradually learn how to focus an object in front of them during the first weeks. Also, during this period of time, babies get used to light and therefore seeing more ranges of shadows. As the retina keeps developing, the ability to recognize patterns improves. At the age of one month, they begin to be sensitive to shades of colour and will look longer at bolder colours and contrasting patterns than at lighter tones. It is by about the age of 4 months that babies can differentiate and respond to the full range and shades of colours. Up to 2 months of age, depth perception and visual coordination constantly improve, which helps them to be able to follow with their eyes an object that is moving. Distance vision also keeps on developing during the early months.

Visual acuity improves quickly between the 1st and the 6h months. Beyond this point, this turns to a gradual improvement. (Table 3.1)

AGE	VA SNELLEN
Birth	6/300
1 month	6/200 – 6/90
3 months	6/90 – 6/60
6 months	6/60 – 6/36
9 months	6/36 – 6/24
12 months	6/24
18 months	6/18 – 6/12
24 months	6/12 – 6/9
36 months	6/9 -6/6
5 years or more	6/6 – 6/5

Table 3.1: The changes in visual acuity from birth with the use of visual preference technique (CET, Continuing, educating and training: "Paediatric Optometry: optometric examination of children"; 2007).

3.3 DEVELOPMENT OF THE REFRACTIVE ERROR

Refraction in normal subjects follow a determinate evolution. The majority of the new-borns present hyperopia, with an average of +2.00D, which increases until the age of 6 months, where it quickly decreases until the age of 2 (table 3.3). Although in new-borns myopia it rarely exists, premature babies may frequently present myopia whereas in these cases myopia tends to reduce to a low hyperopia.

At the age of two, astigmatisms of 1-1.50DC decrease. Emmetropization occurs during the first two years, when it then stabilizes around the age of 3-4 years. The incidence of astigmatism in new-borns without Down syndrome is about 65% greater than 1DC. Infants with strabismus do not demonstrate emmetropization. If there is a refractive error higher than +4D at the age of 6 months, this will tend to reduce to lower hyperopia. However, it can also remain in hyperopia and develop esotropia and amblyopia.

AGE	REFRACTION
3 months	+3.00 D
6 months	+2.50 D
9 months	+2.25 D
12 months	+2.00 D
18 months	+1.50 D
2 years	+1.00 D
3 years	+0.50 D
4 years	+0.50 D
5 years	+0.50 D
6 years	+0.50 D

Table 3.2: Mean refraction from birth as a function of age in children.

3.4 OCULOMOTOR DEVELOPMENT

Oculomotor development also follows a patter in normal subjects. The developing skills of the eye movement system are considerably slower compared to the capabilities of the binocular and accommodative system. Thus, during the firsts 6 months of age the eye movements keep improving, but not until the 1st year of life the binocular and accommodative systems becomes similar to adults. These movements keep developing during the first years of life (Esparza et al; 2014).

Saccades movement of the eyes are defined as ocular movements which allows to fixate on the object we want to observe in a quickly gaze, while pursuits movements are defined as slowly and continued movements which allow to maintain the image of the object in movement, constantly in the fovea.

In contrast, optokinetic nystagmus is an involuntary and uncontrollable movement of the eyes with two phases: one fast, to fix the object, and one slowly, to follow the object and maintain the image in the retina. This type of eye movement is present from birth, in contrast to saccadic and pursuits movements of the eyes, which start to develop by the age of 4-6 weeks to 6-8 weeks (Table 3.2).

MOVEMENT		AGE
Optokinetic nystagmus		At birth
Vestibular ocular reflex		At birth
Saccades	Horizontal	At birth
	Verticals to the top	4-6 weeks
	Verticals to the bottom	3 months
Pursuit		6-8 weeks

Table 3.3: Development of the oculomotor function.

4 CHROMOSOMIC DISORDERS: DOWN SYNDROME

There are many types of chromosome abnormalities. These occur when there is a defect in a chromosome or in the arrangement of the genetic material on the chromosome. There are two main types of chromosomal abnormalities, which can occur during meiosis and fertilization:

- Numerical abnormalities: when the cell results with an extra chromosome or a deficiency in chromosomes. Usually caused by a failure of chromosome division.

- Structural abnormalities: when a chromosome is altered. This occurs due to a loss of genetic material, or a rearrangement in the location of the genetic material. They can be caused by deletions, inversions, duplications, ring formations and translocations.

Most chromosome abnormalities occur as an accident in the egg or sperm. If this is the case, then all chromosomes are affected, however if it happens after conception then some cells have the abnormality and others don't.
Chromosome abnormalities can be inherited from a parent or can be new to the person. That is why it is important to perform chromosome studies in parents (NSGC, "What is Down syndrome?"; 2012)

This study focuses only on chromosome 21 disorder, Down syndrome.

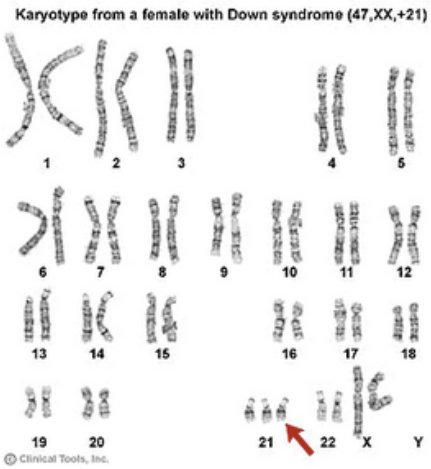


Figure 4.1: Karyotype from a female in which there are 47 chromosomes instead of 46 with Down syndrome (47, XX, +21).

4.1 DOWN SYNDROME

Down syndrome (DS) is a developmental disorder caused by an extra copy of chromosome 21 (fig 4.1), meaning that individuals have three copies of this chromosome. They have 47 chromosomes instead of 46 as normal subjects have. It is one of the most common chromosomal abnormalities in new-borns (Weijerman et al; 2010). Down's syndrome affects 10/10,000 of live births throughout the world (Weijerman et al; 2010).

Johan Langdon Down first described Down syndrome in 1866, but it was not until 1959 that Lejeune et al; (1959) showed that Down syndrome was a genetic condition.

4.1.1 Causes:

The definitive cause for Down syndrome is unknown, but is typically caused by nondisjunction. Nondisjunction happens when a pair of chromosomes don't separate during sperm formation. When the ovule unites with the normal sperm to form an embryo, this ends ups with three copies of chromosome 21 instead of the normal two. The extra chromosome is then copied in every cell while the baby is developing.

Nondisjunctions seem to occur more frequently in older women. This may explain why the risk of having a baby with Down syndrome is higher among mothers of 35 years old and older. Nonetheless, the number of Down syndrome new-borns has recently decreased due to the terminations preferences in older mothers.

In rare cases, Down syndrome can be caused by a Robertsonian translocation. This occurs when the long arm of chromosome 21 breaks off to another chromosome at the centromere. Carriers of this translocation won't suffer Down syndrome themselves, but their children will have.

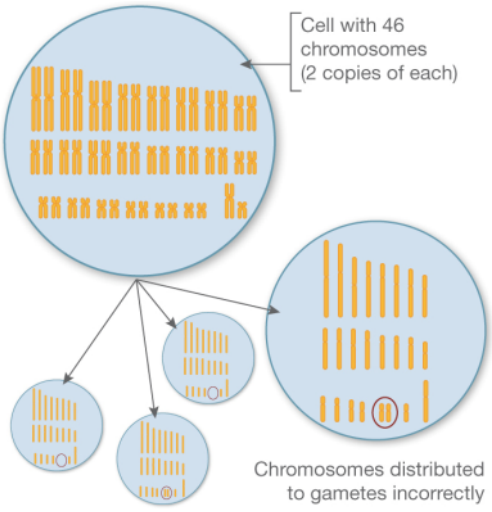


Figure 4.2: Nondisjunction of chromosome 21.

Every person born with this condition is different. There are three kinds of Down syndrome:

- 21 Trisomy (90-95%): characterised by the presence of a complete third chromosome. This occurs as a result of non-disjunction during meiosis.
- Translocation (3-4%): in which one chromosome translocate and attaches to another.
- Mosaicism (1%): when non-disjunction occurs after cell fertilisation resulting in an individual having cells of two types: normal cells and cells of Down syndrome type.

4.1.2 Genetic and medical problems associated to Down syndrome

People with Down syndrome have an increased risk of developing a number of significant medical problems, including: heart disease (approximately 50% of children with Down's syndrome suffer from congenital heart disease (CSDC; 2005), dementia, with an IQ of 20-50 (Randall T. Jose; 1988) hearing and ocular problems.

They may also present thyroid disease and skeleton problems. The respiratory, endocrine (hyperthyroidism) and digestive (12% of new-borns with DS have stenosis) system also seem to be affected. They have poor immunity and are sensible to infections (CSDC; 2005, FEISD)

Subjects with Down syndrome have distinctive facial features. Their typical phenotype has the following characteristics:

- Decreased head circumference with brachycephalic and occipital flattening.
- Facial characteristics: epicanthus, small nose with plane nasal bridge, tongue protrusion, small ears and narrow ear canal and narrow eyes. Strabismus.
- Square shape little hands and feet: Brachydactylic (genetic malformation in which the hand and foot fingers are shorter than usual) and Clinodactyly (angular malformation of the fingers). Single palmar groove and sign sandal, separation between the first and second finger of the foot.
- Skin and appendages: redundant skin in the neck region especially in the neonatal period.
- Low stature and dumpy.

Down syndrome subjects have problems in the development of the speech and difficulties in acquiring abilities to pronounce words, due to their slow development for comprehensive and cognitive skills.

Figure 4.3: Facial physical characteristics of Down syndrome.

4.1.3 Prevalence of Down syndrome

Down syndrome is one of the most common chromosomal abnormalities in new-borns. Throughout the world, the overall prevalence is 10/10,000 births (Weijerman et al.; 2010).

The major risk factor affecting the natural birth prevalence of Down syndrome is maternal age. The older the mothers are, the more of a risk of having a child with an abnormal chromosomal condition. Robles Bello; (2007) showed that in women over 35 years old, mean incidence of having a child with Down syndrome is of 31.3 per 100,000 births, while young mothers have a mean incidence of 1.5 per 100,000. However, over the years, older mothers have more diagnosis tests during pregnancy which leads to an end of the pregnancy term, and so to a reduction of Down syndrome new-borns. Young mothers have fewer incidences.

To a large-extent, the prevalence of Down syndrome depends on several socio-cultural variables. In countries where abort is illegal, such as Ireland and the United Arab Emirates, its prevalence is higher. By contrast, in France, Down syndrome prevalence is lower, and probably due to the pregnancy terminations (Weijerman et al.; 2010, R. E. Stewart et al.; 2007, Robles Bello; 2007).

In the Netherlands, the most recent measure of Down syndrome was 16 per 10,000 live births (Weijerman et al.; 2010). In the United Kingdom, the prevalence of pregnancies affected by Down syndrome has increased significantly, but there has been no overall change in the live birth prevalence of DS (R. E. Stewart et al.; 2007, J. M. Woodhouse et al.; 2000, Kathryn J. Saunders et al.; 1996, M. E. Weijerman et al.; 2010, M. Rosenfield et al.; 2009).

In Europe, Down syndrome accounts for 8% of all registered cases of congenital anomalies. In Spain, the ECEMC (Estudio Colaborativo Español de Malformaciones Congénitas) reported that all chromosomal anomalies diseases have decreased in population, due to abortion after a pre-natal diagnosis. FEISD (Federación Española de instituciones del Síndrome de Down, 2002), reports than round 1/3 of pregnant women in which the foetus presents chromosomal anomalies stop their pregnancy.

In the table 4.1 it can be seen a classification in three time periods of the global incidence of Down syndrome in Spain for every 10,000 new-borns:

	1980-1985			1986-2004			2005		
	Nº	For 10,000	LC 95%	Nº	For 10,000	LC 95%	Nº	For 10,000	LC 95%
DS	565	14.78	13.58-16.02	1822	10.95	10.46-11.46	79	7.40	5.86-9.12

Table 4.1: Incidence and prevalence of Down síndrome for 10,000 subjects (Robles M.A. 2007). LC means de confidence limit.

Going further, the prevalence for every 10,000 new-borns with Down syndrome in the autonomic communities in Spain, from 1980-2005:

SÍNDROME DE DOWN			
	1980-1985	1986-2004	2005
Andalucía	15.37	13.61	5.30
Aragón	-	11.14	0.00
Principado de Asturias	23.32	10.32	23.97
Islas Baleares	4.47	14.10	5.36
Canarias	12.85	7.49	11.88
Cantabria	-	9.98	8.43
Castilla-La Mancha	15.63	12.37	8.87
Castilla y León	14.68	12.42	7.01
Cataluña	16.55	8.25	7.48
Comunidad Valenciana	10.63	7.52	5.94
Extremadura	15.13	10.68	5.44
Galícia	12.63	7.82	2.48
La Rioja	12.55	8.21	0.00
Comunidad de Madrid	16.45	12.80	16.58
Región de Murcia	22.13	12.10	10.35
Comunidad foral de Navarra	14.78	15.92	0.00
País Vasco	13.60	9.30	5.58
Andorra	-	0.00	-
Total	14.78	10.95	7.40

Table 4.2: Incidence and prevalence of Down syndrome among three range of years: 1980-1985, 1986-2004 and 2005 (Robles M.A; 2007).

As it can be seen in table 4.2, the number of new-borns with Down syndrome in Spain has decreased among the recent years, with a prevalence of 7.40 for every 10,000 births in 2005.

The median survival of individuals with Down syndrome has increased considerably in recent years. M. E. Weijerman et al.; (2010) study showed that the median age of death of individuals with Down syndrome has risen significantly in the US, from 25 years in 1983 to 49 years in 1997. In Spain, a 10.8% live over the 45 years of age in 2005 (Robles Bello; 2007).
In the Netherlands, the infant mortality rate in children with Down syndrome dropped from 7.07% in 1992 to 4% in 2003 (M. E. Weijerman et al.; 2010). Congenital heart defects and respiratory infections are the most frequently reported medical disorders on death certificates for individuals with Down's syndrome. Nevertheless, in recent years, the fall in Down syndrome mortality is mainly related to the successful early surgical treatment of CHD (congenital heart diseases) and to the improved treatment of congenital anomalies of the gastrointestinal tract (M. E. Weijerman et al.; 2010).

Since children with Down syndrome have now an improved life expectancy, the total population of individuals with Down syndrome is expected to grow substantially. Preventive programmes for these children will contribute to the improvement of their overall outcome and quality of life.

4.1.4 Ocular findings in Down syndrome

Good vision is important for the development of a child, especially a child with developmental problems such as those associated with Down syndrome as more than half of children with Down's syndrome have ocular abnormalities. These ocular abnormalities include:

COMMON OCULAR ABNORMALITIES	% INCIDENCE	AUTHORS
Accommodating disabilities	50-90%	M. Cregg; 1999, T. Watt et al.; 2014, J.A. Little et al.; 2007, M. Al-Bagdady et al.; 2009
Acquired cataracts	3-15%	M. E. Weijerman et al.; 2010
Amblyopia	20%	
Brushfield spots	38-85%	M. E. Weijerman et al.; 2010, T. Watt et al.; 2014
Blepharitis	7-46%	M. E. Weijerman et al.; 2010, T. Watt et al.; 2014
Congenital cataracts	4-7%	M. E. Weijerman et al.; 2010, A.J. Adams et al.; 1993, B Haargaad et al; 2006, Bhatti TR et al; 2003, Merin S et al; 1971, Wirth MG et al; 2002, Rahi JS et al; 2000
Epicanthic folds	60%	M. E. Weijerman et al.; 2010,
Glaucoma	0.7%	M. E. Weijerman et al.; 2010
Keratoconus	9-18%	M. E. Weijerman et al.; 2010, J.A. Little et al.; 2007
Narrow or Slanted palpebral fissures		M. E. Weijerman et al.; 2010

Nystagmus	11-29%	M. E. Weijerman et al.; 2010, A.J. Adams et al.; 1993, L. Averbuch-Heller et al; 1999, N.R. Bromham et al; 2002
Refractive errors	43-70%	M. E. Weijerman et al.; 2010
Strabismus	20-47%	M. E. Weijerman et al.; 2010, M. Cregg; 1999, T. Watt et al.; 2014, J.A. Little et al.; 2007, A.J. Adams et al.; 1993
Torticollis	9.86%	J. Puig Galy et al.; 2006

Table 4.3. Incidences and literature for the common ocular abnormalities in population with Down syndrome.

Figure 4.4: Ephicantic folds in a girl with Down syndrome.

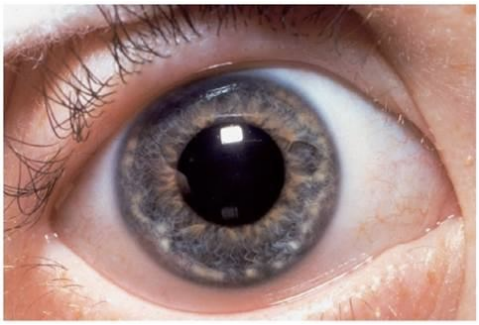


Figure 4.5: Brushfield spots.

Figure 4.6: Esotropia of the left eye in a Down syndrome child.

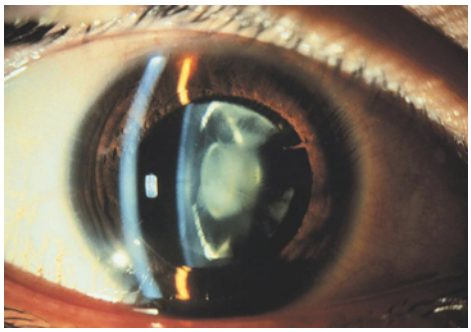


Figure 4.7: Congenital cataract.

Figure 4.8: When Bielschowski is done to the opposite shoulder the vertical deviation appears, caused by the palsy of the superior oblique of the right eye.

5 DOWN SYNDROME VISUAL FUNCTION

5.1 VISUAL ACUITY AND CONTRAST SENSITIVITY

Visual acuity (VA) is reported to have different values for Down's syndrome in comparison to normal age-matched children. During the first years of age, the value is more or less the same between Down's syndrome and normal children, but with increasing age, Down syndrome children tend to have poorer visual acuity than normal subjects. Contrast sensitivity also seems to be generally affected.

Courage et al.; (1994) found that high-contrast grating acuity measured with Teller cards was within normal limits in infants with Down syndrome older than 6 months of age. However, several authors have reported visual acuity decrease when they get older (J.A. Little et al.; 2007, Woodhouse et al.; 1996) who used Cardiff Acuity Test and Teller Acuity cards at three different distances (50cm, 1m, 38cm) to measure visual acuity and compared the results with those of age-matched control children. The study found that VA was well matched with normal subjects in children from early infancy to 2 years of age, but then falls below the normal range with increasing age. This finding applied to children with significant and non-significant refractive error. Haugen et al. (2001) also demonstrated no correlation between refractive error and grating resolution acuity in Down's syndrome. Courage et al.; (1994) also found the same results as Woodhouse et al;. (1996) and Haugen et al.; (2001) using the Teller Acuity cards.
J.A. Little et al.; (2007) measured visual acuity in 16 years old children with Down syndrome. The results found a significant difference between children with Down syndrome and normal subjects. Refractive error and accommodation were not factors in the study as they were wearing their current refractive correction and the test distance was within depth of field. The visual acuity found was 6/13 (0.46 decimal VA) (0.33$\pm$0.18logMAR) for Down syndrome subjects compared to the control group who had VA=6/5 (1.2 decimal VA) (-0.06$\pm$0.07logMAR). Grating resolution acuity in children with Down syndrome was poorer than their age-matched peers. Nonetheless, none of both groups revealed significant association between subject age and resolution acuity.

According to Woodhouse et al. (1996) VA in infants under two years is quite similar to normal children, as it is the spread of the refractive errors, while in children over two years of age, acuity is worst in children with Down syndrome and the spread of refractive errors is significantly different between both groups. However, VA was measured with their habitual correction and not with the refraction found in the eye examination, which could be an influence in the results. Low accommodative response to the close viewing distances used for some measurement could have reduced the performance.

In conclusion (Little et al.; 2007 and Woodhouse et al.; 1996), taking the cognitive abilities of the child into account, there are normal levels of vision before six months of age, but visual acuity decreases when DS children get older.

Regarding contrast sensitivity, Courage et al. (1997) measured across five spatial frequencies in Down syndrome with ages ranging from 4 months to 14 years. The results for the Contrast Sensitivity Function (CSF) curve were at the lower limits, but within the 90% interval of the normal range.

These two figures (5.1, 5.2) show how Contrasts sensitivity function (CSFs) fall below within the normal limits:

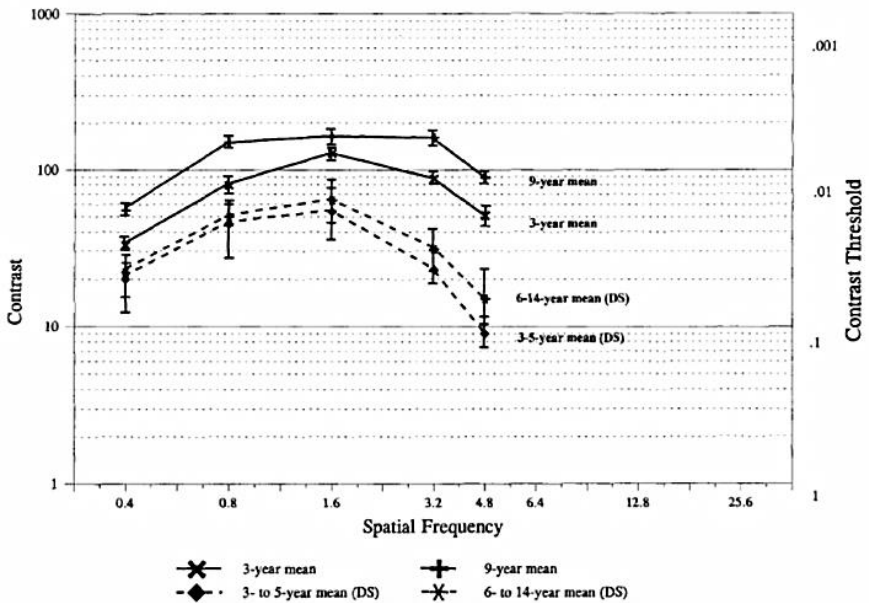


Figure 5.1: Mean contrast sensitivity function (with standard errors) for 3-5 years olds and 6-14-years-olds with Down syndrome. Mean CSFs (with standard errors) from 3- and 9-years-olds without Down syndrome are also shown for comparison (M. L. Courage et al.; 1997).

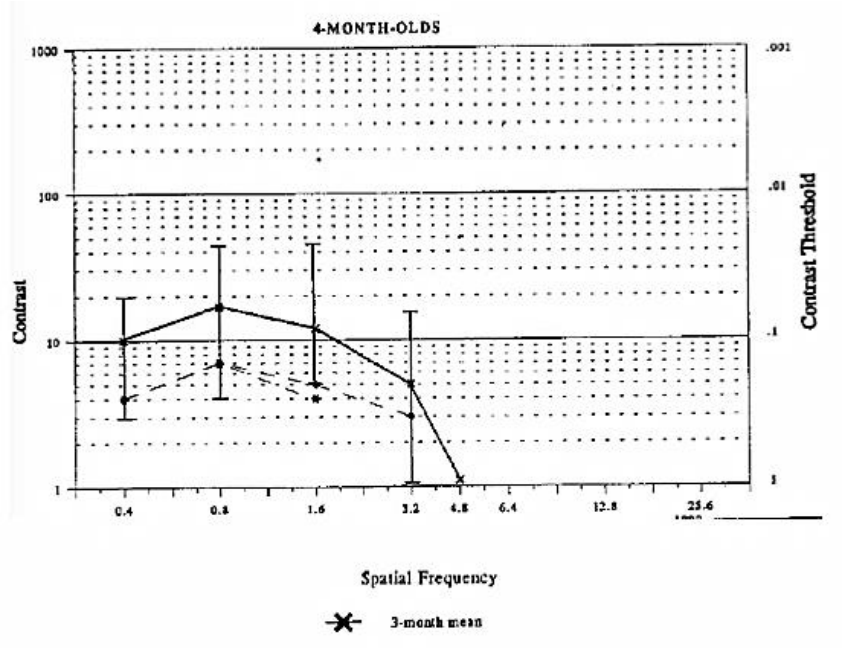


Figure 5.2: Comparison to the normal range of CSFs reported for 3-month-old infants without Down syndrome (M. L. Courage et al.; 1997).

Figure 5.1, shows clearly that despite the same general shape, the mean CSFs of the children with Down syndrome are depressed, especially at higher frequencies.
Although the study was done in a small sample size and this limits any formal statistical comparison, it appears that there is little developmental improvement in CS from the younger to older groups.
It can also be seen that there is a little overlap between the CSFs of the 3- and 9-year-olds without Down's syndrome, and this difference is highly significant (Adams & Courage; 1996*).*
Figure 5.2. reveals that the general shape of the CSFs of the two infants with Down syndrome is similar to that of the 3-month old mean. However, although within the normal range, it is also clear that their CSFs fall consistently in the lower part of the range across all spatial frequencies tested (M. L. Courage et al.; 1997).

Courage's finding was a greater difference between children with Down syndrome and non Down syndrome at higher spatial frequencies (finer gratings). The general shape for CSF was quite similar between the children with Down syndrome and normal children, although the mean CSF of children with Down syndrome at 7.3 years old was similar to the CSF of a normal infant of 12 months old.

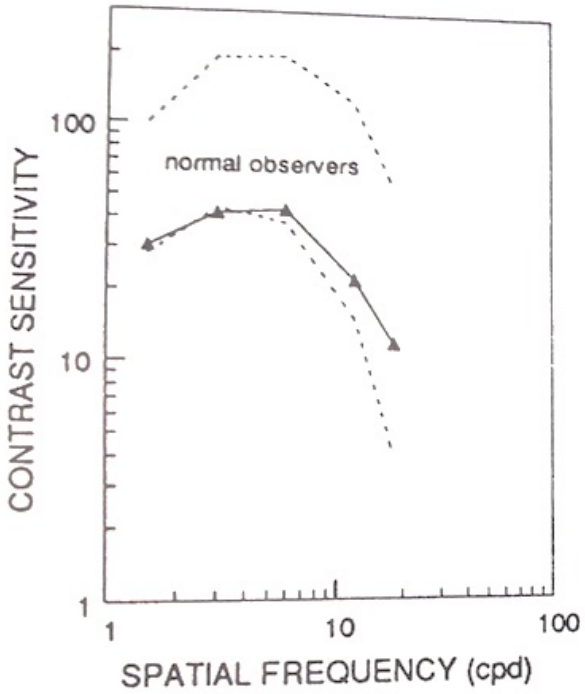


Figure 5.3a.

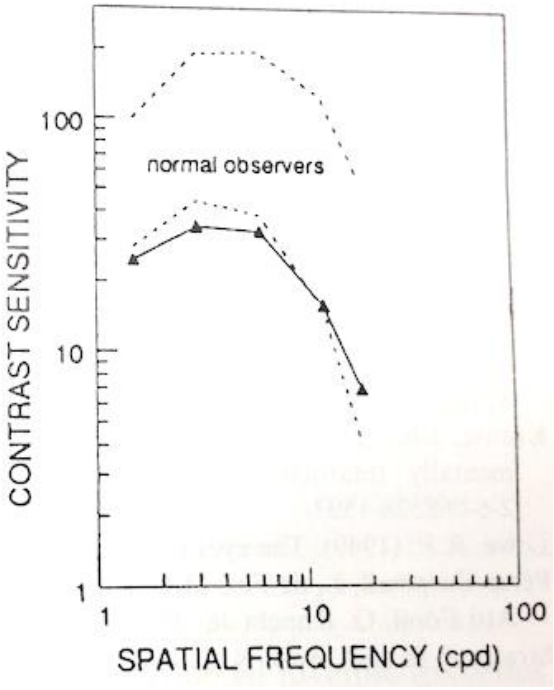


Figure 5.3.b.

Graphic 5.3a. and 5.3b. show contrast sensitivity moderately retarded (A) and severely retarded (B) in Down syndrome subjects. The dotted lines show the limits for 90% of the normal population (M. Cregg; 1999).

To sum up, contrast sensitivity is found under the normal limits for Down syndrome subjects in comparison to normal subjects, although it follows the same general shape (figure 5.1 and 5.2). However, figure 5.3a and 5.3b demonstrate that contrast sensitivity in subjects with severe mental retardation is greater compared to subjects with moderate mental retardation, as would be Down syndrome.

5.1.1 Reasons for poor Visual Acuity and Contrast sensitivity:

The aetiology for this poor visual acuity is not fully understood. J.A. Little et al. (2007) found that visual acuity thresholds were significantly poorer in the Down's syndrome subjects in VEP and behavioural measures compared to normal subjects.
Poor VEP performance and visual deficits in Down's syndrome could not be attributed to motivation and attention, as a real sensory deficit exists.

Differences in the visual cortices have been reported (F. M. John et al.; 2004 and M. E. Weijerman et al.; 2010) in subjects with Down syndrome. These include lesser brain weights, less organized configuration of layers in the visual cortex and dendritic atrophy and poor maturation. However, Ellingston (1986) demonstrated only mild and

transient differences in flash VEPs in new-born's with and without Down's syndrome that disappeared after 6 months of age.

It is suggested that the visual deficit in Down's syndrome may occur at any point in the visual pathway up to the primary visual cortex, but not located in the higher cortical areas. Optical, retinal and neural factors may all be implicated, but not attention and motivational factors. Watt et al.; (2014) give evidence that decreased attention is not an explanation of their decreased visual performance. If the acuity loss for children with Down's syndrome had been due solely to decreased attention, it would be expected to appear a bigger difference between behavioural acuity relative to VEP acuity for children with Down's syndrome compared to normal subjects.
John F.M et al.; (2004) used steady state visually evoked potentials (VEPs) to measure acuity and contrast sensitivity and compared children with Down syndrome and age-matched children, excluding those with inaccurate accommodation.

The optical components of the eye in Down syndrome are known to be increased risk for abnormalities including early-onset cataract, keratoconus, refractive error and poor accommodative function. Poor optical quality has implications for retinal and cortical image quality (J.A. Little et al.; 2007).
Other studies have described the retinal structure in Down's syndrome. These studies report retinal anomalies or retinal abnormalities, mostly associated to high myopias or incidences of retinal detachments (Ahmad et al.; 1976, Roizen NJ et al.; 1994, Da Cunha RP et al.; 1996, Berk AT et al.; 1996, Liza-Sharmini AT et al.; 2006) and cite instances of optic disc elevation, macular coloboma, congenital amaurosis, possible link between Down syndrome and retinoblasma, and increased vasculature at the optic nerve head (J.A. Little et al.; 2007).
Structural evidence exists that the crystalline lens is thinner in persons with Down syndrome (Haugen et al.; 2001). Decreased central corneal thickness (Haugen et al.; 2001, Everekliogl u C et al.; 2002) and increased keratometry indicates steeper corneas (Haugen et al.; 2001, Vincent AI et al.; 2005). Differences in tear film integrity have been reported with reduced numbers of goblet cells in the bulbar conjunctiva (Filippello M et al.; 1997). This can also be a factor for which visual acuity is affected in Down syndrome.

Children with Down syndrome had a small but statistically significant reduction in interferometry acuity compared with normal subjects. Interferometric acuity was 6/6 in Down syndrome children compared to normal children 6/4.7 (1.27 decimal VA). This small loss of interferometric acuity in children with Down's syndrome compared to the normal subjects does indicate the possibility that there is a small post-retinal component to the reduction in vision in Down's syndrome.
Pre-retinal contributions to a reduction in visual acuity could be due to visually significant cataract or keratoconus. However, Courage et al. (1997) found that despite lower contrast sensitivity seen in children with Down syndrome none had significant cataracts nor keratoconus and all had less than 1.75DC astigmatism (T. Watt et al.; 2014). This low contrast sensitivity was particularly seen at lower spatial frequencies.

A decrease in the accuracy of accommodative response might explain the reduction in CSF at high spatial frequency, but this cannot explain the generalized decrease at lower spatial frequencies observed in the CSF, which suggests that the least part of the visual deficit seen is cortical. Suttle and Tuner (2004) made objective measurements of the cortical visual response and the results were less clear for Down's syndrome children than normal children, adding evidence to the hypothesis that there is neural basis for the deficits in contrast sensitivity.

5.2 REFRACTIVE ERROR

5.2.1 Spherical Equivalence Refraction (SER):

Among people with Down syndrome, significant refractive errors occurs far more commonly than among the general population. In normal developing children, significant refractive errors are common in the first year of life. Cross-sectional data (M. Al-Bagdady et al.; 2010, T. Watt et al.; 2014) showed a considerable difference between the two populations.

	INFANTS	PRESCHOOL	PRIMARY SCHOOL
Down syndrome	30%	50%	54%
Controls	25%	5.8%	3.2%

Table 5.1: Prevalence of significant error at three levels of development. (T. Watt et al.; 2014)

In new-borns, spherical refraction is normally distributed with a mean of low hyperopia. Then, the refractive errors decline during the first few years of life, so that very few children have refractive errors by early school age. This process of decrease of refraction power is termed emmetropization (J. M. Woodhouse et al.; 1997, M. Cregg et al.; 2001). The range of refractive errors widens with age in the first four years of age and remains wider (M. Al-Bagdady et al.; 2010).

During infancy and early childhood, children with and without Down syndrome tend to be hyperopic with a wide distribution in refractive errors. Nevertheless, throughout childhood and teenage years, in normal subjects the prevalence of refractive errors decrease shifting from high hyperopia to low hyperopia/myopia, while in Down's syndrome refractive errors are fairly stable. Myopia can start to develop in Down's syndrome when they are older (M. Al-Bagdady et al.; 2010). This increase in variability of refractive error is proposed to occur because of a failure of emmetropization.

5.2.1.1 Reasons for significant refractive error in Down's syndrome:

Olav H Haugen et al. (2001) indicates that reduced accommodation in early age, causing a blurred retinal image for objects at near, may be of aetiological importance for the abnormal refractive development. Obviously, there must be additional factors.

For example, reduced accommodation would shift the optical focus behind the retina thus induce a myopic shift. However, this does not explain the cases with increasing hyperopia.

It is also reported that the failure to a decrease of refractive error is also due to a failure of emmetropization. A possible explanation for this failure includes:

- Inaccurate accommodative response
- Low levels of near work combined with high levels of outdoor activity
- Changes in the visual cortex

(Olav H Haugen et al, 2001)

Nevertheless, as said before, hyperopia has a higher prevalence than myopia so accommodation cannot explain the failure of emmetropization (T. Watt et al.; 2014).
There is evidence that outdoor activity and near work can influence a child's refractive error. A low level of near work combined with high number of hours outdoor is associated to more hyperopic spherical equivalent refractive error. Nonetheless, this argument is weak (Tanisha et al.; 2014).
In conclusion, further research is needed to determine why emmetropization fails in children with Down's syndrome.

5.2.1.2 Refraction results comparing Down's syndrome and age matched normal subjects:

The longitudinal study from J. M. Woodhouse et al.; (1993) showed that primary school children with Down syndrome have higher prevalence of significant refractive error (>-0'75 or >+3'00D) compared to normal. They found a median in refractive error of +1D among subjects with Down syndrome, while children with no Down syndrome had a media of +0.75D. They also found that the range of refractive errors among subjects with Down syndrome was from -12D to +3.5D, whereas normal subjects ranged from -0.75D to +3.75D. Also, the prevalence of significant refractive error increased over time and that spherical equivalence refraction in Down syndrome primary school children had a larger variability than normal subjects (J. M. Woodhouse et al.; 1997 and T. Watt et al.; 2014).

J. M. Woodhouse et al.; (1997) found that children with Down syndrome during their first year of age, 30.40% had significant refractive errors, 9% of which were myopic and 21% hyperopic, whereas in the normal children only 25% had significant refractive error and all of them were hyperopic. Wesson M.D; (1995) suggests no change in mean refractive error for children with Down syndrome aged between 12-84 months. T. Watt et al.; (2014) found not much difference between both groups in For preschool children their finding was that 50.6% children with Down syndrome had significant refractive errors, 8.8% of those were myopic and 41.8% hyperopic (T. Watt et al.; 2014, J. Puig et al., 2002). However, among normal subjects 5.8% had significant errors, all of them hyperopic. Finally, 54.6% of children in primary school children with Down syndrome had significant error, of whom 12.6% were myopic and 42% were hyperopic.

In contrast, 3.2% of the normal children had a refractive error (1 out 31 subjects examined) and all of them were myopic. As it can be seen, after infancy, the difference in the prevalence of refractive error between those subjects who suffer from Down's syndrome and those who don't is greater (J. M. Woodhouse et al.; 1997).
J. Puig et al. (2002) study, 72% cases of children with Down syndrome were emmetrope or hyperopic and 28% were myopic. Myopia becomes more common up to adolescence.

Type of refractive error	Frequency (%)
Hyperopia	
≥+1.00 DS	56
>+2.00 DS	57
≥+0.50 DS	80
Myopia	
≤-0.50 DS	18–25
<-0.50 DS	12
Emmetropia	
Between -0.50 and +1.00 DS	19
Between or equal to -0.50 and +2.00 DS	32
Between or equal to -0.50 and +0.50 DS	2
Astigmatism ≥ 1.00 DC	67–74
Anisometropia ≥ 1.00 DS	9–19

Figure 5.4: The prevalence of different types of refractive error in children with Down syndrome (T. Watt et al.; 2014).

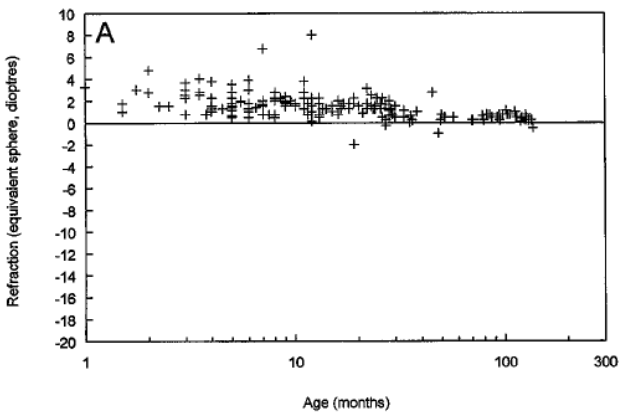


Figure 5.5a.

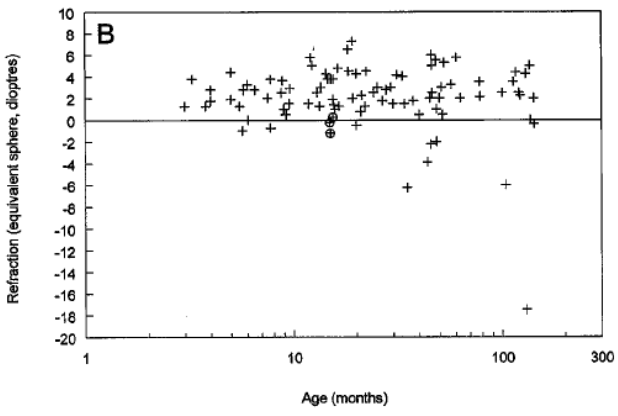


Figure 5.5b

Figure 5.5: Refraction expressed as equivalent sphere in dioptres, for the right eye. 5.5a) In normal subjects. 5.5b) In children with Down syndrome. Data points for the three children with Nystagmus are circled.

As figures 5.5a and 5.5b show, in normal subjects the refractive error is present at the age of birth and declines during the firsts months of age until it disappears by the age of 3 years (table 3.3, figure 2.a) while in Down syndrome subjects this does not happen and the refractive errors remains with ageing. Both figures show how emmetropization happens in normal children but not in Down syndrome. It can also be seen that hyperopia is the most common type of refractive error compared to myopia the spread of power value is wider for Down's syndrome than in normal subjects (figure 5.5a, 5.5b).

5.2.2 Astigmatism:

Differences between the children with Down syndrome and normal subjects are also apparent with astigmatism. Astigmatism follows a similar pattern of refractive error with normal subjects, being common in early infancy and prevalence declining along with spherical errors in childhood (J. M. Woodhouse et al.; 1997). However, astigmatism in Down's syndrome doesn't seem to decrease as there is no emmetropization.

5.2.2.1 Reasons for astigmatism in Down syndrome:

The increasing incidence and rise in the power of astigmatism may be caused by the effect of the eyelids on the cornea, as they have an obliquely slanted palpebral fissure and thinner corneas. It has been proved (M. Al-Bagdady et al.; 2010) that the axis of the oblique astigmatism is correlated with the slanting of the palpebral fissure.

A working hypothesis for the development of astigmatism is that eyelid pressure at an oblique angle could result in oblique astigmatism and that the increased magnitude of astigmatism represents an accumulation of flattening by the mechanical force of the eyelid. Read SA et al; (2007) and Shapiro MB et al.; (1985) found that corneal astigmatism power vector was significantly correlated to the angle of the palpebral fissure.

It is also said that the increase of astigmatism over time could be due to the development of keratoconus, which is found in up to 15% of people with Down syndrome, although Woodhouse J.M.; (1997) found that of 37.5% of primary school children with Down syndrome who had 1.00DC or more astigmatism, none had keratoconus. So this is not a strong enough evidence to say they are related with each other. However, keratoconus develops around puberty, so it may be the reason why none of those children had keratoconus.

The change of astigmatism over time is a strong reason for having a regular eye examination. Non-correction of significant astigmatism at an oblique angle will reduce vision and will increase potential for refractive amblyopia (T. Watt et al.; 2014).

5.2.2.2 Astigmatism results comparing Down's syndrome and age matched normal subjects:

Astigmatism decreases with age in children with no Down syndrome, so that by primary school age very few children have considerable astigmatic error. This doesn't happen to children with Down syndrome. During infancy, the prevalence of astigmatism appears to be lower in Down's syndrome group than in normal children. However, astigmatism does not disappear from the Down's syndrome population at older age.

Some studies have proved that with-the-rule is the major type of astigmatism in children with Down syndrome during his infancy, while children without Down syndrome tend to have against-the-rule astigmatism. Afterwards, oblique astigmatism seems to appear in Down's syndrome while for normal subjects the emmetropization seems to happen and astigmatism tend to disappear (M. Al-Bagdady et al.; 2010).

In all the studies, astigmatism greater than 1.00DC is considered severe astigmatism. Ljubic A et al.; (2011) found that astigmatism greater than 1.00DC was present in 74% of people with Down syndrome aged between 1-34 years old and oblique astigmatism was the most prevalent type (52%). Other studies such as Al-Bagdady (2011) and Haugen O.H.; (2001) made the same observations. They found that astigmatism first became significantly different in 7 years old children compared to 2-year-old children. In their longitudinal study 7 out of 12 children developed significant astigmatism and 6 out of 7 had oblique astigmatism. Puig et al.; (2002), 17% of the children had astigmatism equal or superior to 2.00DC.

J. M. Woodhouse et al.; (1997) found that among infants with Down syndrome, 26% had astigmatism of 1D or greater in each eye, while in normal subjects the prevalence of astigmatism was 48.1%. Among preschool children, Down's syndrome demonstrated a prevalence of 22.2-30% of astigmatism of 1D or greater (J. Puig et al., 2002, J. M. Woodhouse et al.; 1997, Olav H Haugen et al.; 2001), while non Down syndrome children had an incidence of astigmatism of 15.4%. In primary school children, 37.5% of Down's syndrome had astigmatism. In contrast, normal children there were no cases of significant astigmatism (J. M. Woodhouse et al.; 1997). Bittles AH et al. (2004), Kleinstein RN et al. (2003), Deng L et al. (2012) also concluded that for infants and preschool children, the distribution of astigmatism does not differ significantly between children with Down's syndrome and non Down's syndrome. Ljubic A et al. (2011) also obtained similar results. He found that the frequency of significant astigmatism increased with age for Down's syndrome children. In infants, 26% had significant astigmatism compared to 22% of the same children at preschool age and 37.5% at primary school. This contrasts with the trend of a reduction in

astigmatism in subjects with non Down's syndrome. About 48% of normal subjects had significant astigmatism in infancy age, 15.4% at preschool age and 0% at primary school age.

The study of M. Al-Bagdady et al. (2010) and T. Watt et al. (2014) showed a major prevalence of with-the-rule astigmatism in Down's syndrome children during early childhood, which seem to disappear by the age of 4 years. However, with time these subjects started to develop oblique astigmatism. It was observed in those subjects with Down syndrome who had oblique astigmatism a change in the axis of the cylinder. From the 14 eyes out of 15 in which astigmatism was measured, showed the same right-left pattern with an axis of 135º in the right eye and 45º in the left eye. It is suggested that specific direction is caused by the upward slanting of the palpebral fissure (T. Watt et al. 2014, Olav H Haugen et al. 2001 and Al-Bagdady, 2011). The pressure from the eyelids has already been pointed out as a major aetiological factor of corneal astigmatism (5.6.1).

5.2.3 Anisometropia:

The reported prevalence of anisometropia in children with Down syndrome is significantly greater than in the general population as a difference in refraction between the two eyes of 1D or greater is not common among typically developing children. (T. Watt et al.; 2014). Nevertheless, it is found great difference in the prevalence of anisometropia in primary school. J. M. Woodhouse et al. (1997), M. Al-Bagdady et al. (2009), all found in normal children an incidence of anisometropia of 8.6% in infants, 4.6% in preschool and 3.2% in primary school children, while in Down syndrome children the prevalence was of 4.3% in infants, 13.3% in preschool children and 20.8% in primary school.

5.2.4 Treatment:

Spectacles are the most common treatment for refractive errors in subjects with Down syndrome. Contact lenses could be also used as a treatment, although the subject should be aware of the requirements and the cares needed. Lasik surgery could also be used as a treatment, though it is not recommended in young subjects.

Prescribing specs for hyperopias have shown that it doesn't improve near focusing, and those with myopia may be worse off when wearing glasses (M. Al-Bagdady et al.; 2010). The problem that myopic children have to face is that they would be able to see near targets clearly, however they would see distance targets blurred. When corrected, if the accommodation is poor, then there is blurred vision in near. So, one of the possible factor for rejecting wearing specs is because they cannot see at near.

To persuade a person to wear glasses can be difficult at the beginning. So it is of utmost importance that the glasses fit comfortably that will avoid the child try to remove them if they are digging into her face or that back of the ear. Moreover, it is

important that the child gets used as soon as possible to wear the glasses, so if the child enjoys doing an activity and does it with the glasses, he will get the routine quickly and get used to it, associating the glasses with enjoyable activities.

5.3 ACCOMMODATION:

Some studies have reported that Down's syndrome have reduced accommodation which is even present when infants are three months of age. The amplitude of accommodation also seems to decrease with age and give rise to presbyopia symptoms, although is not presbyopia.

Under-accommodation may have implications for the defective visual development of these children. Visual acuity does not reach normal levels, and contrast sensitivity may be reduced from an early age. A consistently blurred retinal image may be crucial in the onset of these deficits (M. Cregg et al.; 2001, Karlica D et al.; 2011).

The greatest difference found between Down syndrome children and typical children is at near vision. Normally, children focus very well and accurately on near targets, and as people age, this ability to focus at near is weakened. Over 70% of Down syndrome children don't focus properly at near and they tend to under-accommodate (J. M. Woodhouse; 2005).

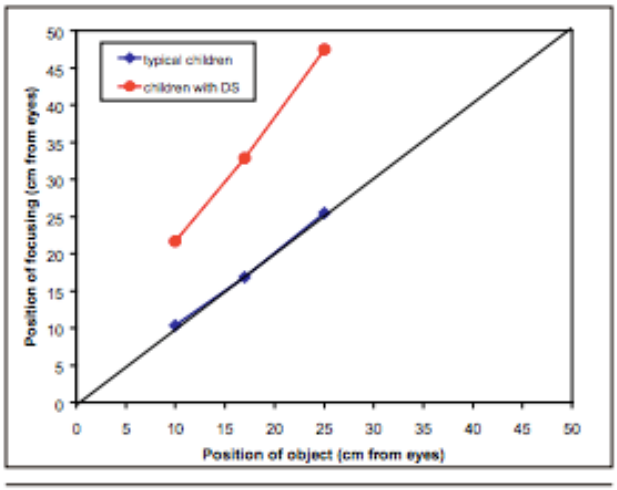


Figure 5.6: Near focusing in children with and without Down syndrome.

This is consistent for any individual child, and persists even when children is corrected with their spectacles for distance.

The limit of accommodation in Down syndrome children is 6-8D, equivalent to 16-12.5cm , so closer targets cannot be presented as they won't be able to see them clearly.

M. Cregg et al.; (2001) used the dynamic retinoscopy technique, but with a dynamic target at 6D distance. The position of neutralization was obtained by the examiner moving away from or towards the subject. The target was moved then to distances equivalent to 8-11D (12.5-9cm) and the position of neutralization was noted again.

The validation they used for this exam was:

- Accuracy of accommodation: significant difference between the accuracy of accommodative response of the adult subjects and the children at all three distances.
- Amplitude of accommodation: is the maximum accommodation that a subject can exert. In children, the nearest target distance used was 11D (closest target could be held). The older adult group it was considered a considerable lag at near target distance (1-6D).

In general, young children with no Down syndrome show accurate accommodation to targets at 4-6D (25-16cm) and a lag of accommodation of more than 1D is considered abnormal. The normal values of accommodation in children without the syndrome in 6 years old is 11.94-18.87D amplitude (8.3-5.3cm), at 10 years old between 13.56-18.94D (7.3-5.2cm) and 10D (10cm) is the lowest amplitude that would be considered normal in children of this age range. A research from Rouse et al.; (1984) found the usual mean lag of accommodation in normal subjects was 0.3D (0.33m) and the average working distance was 24.6cm (4D). Nonetheless, this is different for children with Down syndrome. Rouse et al.; (1984) found that 55% of Down syndrome children had a lag of accommodation grater than 1D at working distances between 20-30cm.

Woodhouse et al.; (1993) measured the amplitude of accommodation using Nott's dynamic retinoscopy at three different distances. Refractive error was not corrected. The results showed that 50% of Down syndrome children had amplitude of accommodation of 4D (25cm) or less. In normal subjects, the amplitude of accommodation was less than 10D only for 7.6% compared with the 92% of the children with Down syndrome. Those with Down syndrome have mean amplitude of accommodation 2.52 ± 1.66D (Anderson HA et al.; 2011).

M. Cregg et al.; (2001) study demonstrated a large under-accommodating at all distances tested, for Down's syndrome. In comparison to subjects with no Down syndrome, in which a lag of less than 1D at 10cm was present, Down's syndrome subjects may show a lag of accommodation as large as 5D for a target at 10cm from child's eyes. This lag of accommodation is present even when infants are three months of age. So, 92% of Down syndrome children had an amplitude poorer than normal value, 80% had reduced accommodation and 50% had amplitude of 4D or less. Also, in study of Olav H Haugen et al; (2001), 50% of Down syndrome children had accommodation weakness. A normal adult subject with these values would require a presbyopic spectacle correction which would be considered essential.

5.3.1 Reasons for reduced accommodation:

The reason for the poor near focusing is not still know, however some studies have considered the crystalline lens mechanics setting limits on the total accommodative as a possible explanation for the lower amplitude of accommodation, as it happens in presbyopia (M. Cregg et al.; 2001). Yet, it had to be discarded as there is no evidence of saturation of accommodative response at the targets. Another point in defence that DS are not presbyopes is that presbyopia shows the same result of accommodation in all near targets. In contrast, all children with Down's syndrome who under-accommodated for near targets in this research showed lag of accommodation that varies consistently with target distances. It is important to be aware that as refractive error changed, so did the total accommodative response (M. Cregg et al.; 2001).

Some studies found that the central lens was thinner on average in people with Down syndrome compared to normal subjects (3.27 ± 0.29mm in DS and 3.49 ± 0.20mm in normal subjects) (Haugen OH et al.; 2001). Also, the lens power has been calculated and was significantly lower in Down's syndrome (12.70 ± 2.36D) compared to controls (19.48 ± 1.24D).
Moreover, the accommodation and vergence neural control mechanism in these subjects could be different. This involves a number of areas in the brain that needs to be more investigated.

Another suggested explanation for the reduced accommodation of children with Down's syndrome was the pupil size. A small pupil is consistent with a large depth of focus. The depth of focus of any eye is dependent partly on pupil size. A smaller pupil gives rise to a larger depth of focus. However, M. Cregg et al.; (2001) found that all subjects presented normal pupil size, so under-accommodation cannot be related directly to the pupil size.

It also appears that the accommodation system of the children with Down syndrome may have the physical capacity to respond to a given stimulus, but the neural control of the system is defective. The system appears to be well regulated, as evidenced by the consistency of the accommodative response in any individual child.

Haugen and HØvding; (2001) suggested that a weak accommodation might be caused by a general malformation of the parasympathetic nervous systems, including the enzymes choline acetyltransferase and acetylcholinesterase. Nonetheless, this needs further research.

5.4 DISORDERS OF BINOCULAR VISION

5.4.1 Strabismus:

A high prevalence of strabismus (20-47%) has been reported in children with Down syndrome (J. Margaret Woodhouse et al.; 1997, Gardiner PA; 1967, Millis E.A.; 1985, T. Watt et al.; 2014, Olav H. H.; 2001).

5.4.1.1 Causes for strabismus in Down syndrome:

The causes for infantile strabismus are not clear, but it is known that the presence of strabismus in Down syndrome subjects may be predictive of a significant refractive error. Nonetheless, the association between strabismus and the process of emmetropization is also unclear.

Although accommodative and refractive esotropia is expected to be more common in a population with a high prevalence of hyperopia when this is not corrected, if accommodation is reduced then this cause is less likely to occur (T. Watt et al.; 2014). Despite this fact, Ljubic A. et al.; (2011) found more or less the same percentage of cases with esotropia in hyperopic and myopic Down syndrome children (28% and 40%) respectively.

The high prevalence of accommodative insufficiency may be explained by the accommodative-vergence system. As it has been mentioned in section 5.3, Down syndrome subjects commonly display under-accommodation. The increased accommodative effort when trying to compensate for the accommodative weakness presumably precipitates an esotropia at near. So, esotropias are more expected to be found in Down syndrome children than exotropias.
However, accommodation weakness is also common in children without Down syndrome, so there must be additional contributing factors to the cause for strabismus. Olav H. H.; (2001) and T. Watt et al.; (2014) suggested a weak fusion capacity, but this needs further investigation.

Olav H. H.; (2001) suggested that brain damage may be a contributing factor in concomitant strabismus in Down's syndrome. Exodeviations are rare, however, in case of brain damage exodeviations are more likely to occur. This contradicts that brain damage should be a major etiological factor in exodeviations in this subjects as esotropias are the most common type of strabismus in this subjects. Moreover, if accommodative weakness is the major cause for esotropia in Down's syndrome, then it is not known whether this is due to a central or a peripheral defect.

It has been also observed that in some cases the child alternates between squinting with the right eye and the left eye. In others, the child squints constantly with the same eye. The non-correction of the refractive error may lead to appear a strabismus.

Often it is more difficult to recognise a strabismus in children with Down syndrome because of the distinctive appearance of the eyelids. For this reason, it is desirable for all children with Down syndrome to have additional routine screening as recommended in the Down's syndrome Medical Interest Group Health Check Guidelines and J. M. Woodhouse et al.; (1997).

5.4.1.2 Literature review of strabismus in subjects with Down syndrome:

Olav H. H.; (2001) made a cross-sectional data investigation in order to examine the frequency of early strabismus from Down syndrome children during their first year of life. Hirschberg's corneal reflex method was used to determine the eye alignment in near vision.

They defined esotropia, exotropia or vertical deviations as a deviation from the straight position. They also observed if patient had unilateral or alternating strabismus and if they were manifest, intermittent or latent. Infantile esotropia was defined as a constant esotropia with an onset before one month of age. All other cases of esotropia were classified as acquired.

Their findings were in line with other studies about the presence of strabismus in Down syndrome children. They found that 42% of the children examined suffered from strabismus. J. M. Woodhouse et al. (1997), T. Watt et al. (2014) showed a prevalence for strabismus around 19-34% (T. Watt et al.; 2014), 45-50% (J. M. Woodhouse et al.; 1997) and 44% (J. Puig et al., 2002). They also found that from those children with Down syndrome who suffered from strabismus, 84% had esotropia. The cross-sectional study that Olav H. H.; (2001) also proved that the frequency of infantile esotropia in Down's syndrome is similar to that of a normal population (1-2%). In the same study, the predominant type of strabismus in Down syndrome children was an acquired esotropia, affecting 1 out of 3 subjects of the sample with usually an onset from 3 to 6 years old. Most of the cases were associated to hyperopia, which was also combined with an accommodation weakness. In most cases, 75% of the subjects with Down syndrome and acquired esotropia had significant hyperopia at the last eye examination, with mean hyperopia $\pm$4.3 $\pm$1.7D. Good binocular sensory function was found in 40% of all the children with acquired esotropia. In contrast, in normal individuals, infantile esotropia is more frequent than the acquired forms.

The frequency of all types of strabismus did not differ significantly according to the longitudinal refractive development. However, in the group with lower hypermetropia, esotropia occurred less frequently.

Haugen O.H.; (2001) showed that hyperopia was greater in those subjects with Down syndrome who had strabismus (46%) than in those who did not (13%)(figure 5.6). Besides, it was found that the 75% of those who had more than 4D of hyperopia were strabismic, but 22% of children who had less than 2D of hyperopia had strabismus too. Data also showed that when esotropia is present, it is more likely to be alternating (70%) than unilateral (30%), but also that in long-term research a 42% of the children

with Down syndrome who did not suffer from strabismus developed strabismus by the end of the research.

Da Cunha R.P.; (1996) found that non-accommodative and accommodative esotropia were equally common and attributed the vertical deviations to congenital fourth palsy and double elevator palsy.

Although it is not really known the relationship between emmetropization and strabismus, one of the hypothesis for the late onset of strabismus in children with chromosomal anomalies and with uncorrected hyperopia and accurate accommodation is that strabismus may be linked to the potential failure of emmetropization and accommodative esotropia (T. Watt et al.; 2014). For example, Da Cunha RP.; (1996) found that in normal children, the age of onset of accommodative esotropia was between the 2-3 years old. By contrast, in children with Down syndrome, the age of onset of accommodative esotropia was 4.5 years of age. They suggested that this late onset might be caused by a developmental delay and to the increased incidence of high refractive error, which changes with time.

It has also been reported that the high incidence of strabismus goes hand-in-hand with the development of amblyopia. Ljubic A et al.; (2011) reported the frequency of amblyopia to be 17% in children and young adults with Down syndrome between 1 and 34 years old.

The prevalence of strabismus varies with age among Down syndrome children.
J. M. Woodhouse et al. (1997) found different incidences of strabismus in children with Down syndrome. In her study 9% of the infants had strabismus, while preschool and primary school children had 20% and 25% respectively. All 17 strabismus cases were esotropic, with five of them intermittent and with the remainder constant or alternating. In the preschool group, all 9 strabismus children had a significant error, 6 were hyperopic and 3 were myopic. Among the primary school children, 4 had significant hyperopia and 1 had significant myopia and the last one had no significant refractive error. They also found that some children who had a significant refractive error and did not have strabismus (J. M. Woodhouse et al.; 1997).

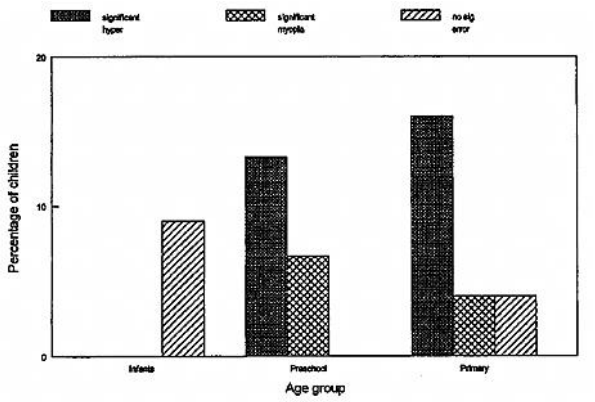


Figure 5.7.:Prevalence of strabismus among children with Down syndrome, divided into those children with a significant refractive error (hyperopia of +3.00D or greater, and myopia of -0.75D or greater) and those without. (J. M. Woodhouse et al.; 1997)

Figure 5.7 shows the incidence of strabismus depending on the type of refractive error divided into the three ranges of ages: infancy, preschool and primary school children. As it can be seen, hyperopes tend to have more incidence for strabismus than myopes or subjects with no refractive error. This happens for preschool children and either primary school children.

J. Puig et al.; (2002) found that esotropia, esotropia associated to vertical deviation, exotropia and isolated vertical deviation were present among the 44% of the subjects with Down syndrome who had strabismus, being esotropia the most common strabismus and vertical deviation strabismus the least.

The vertical deviations they found in the subjects related to horizontal deviation were because of a superior oblique paralysis, a hyperaction of superior oblique, an hyperaction of inferior oblique or a deficit in elevation in adduction, compatible with Brown syndrome[1]. Vertical deviation was caused by an elevation paralysis, or an inferior rectus paralysis, a superior oblique hyperaction or by an inferior oblique paralysis, in which the last one was the most commonly found. 7.08% of those Down syndrome children who suffered from strabismus also had torticollis secondary to their ocular pathology. The torticollis found was with elevated chin or either head inclined to one shoulder.

[1] Brown syndrome also known as Oblique Tendon Sheath syndrome it is a mechanical problem in which the superior oblique muscle is unable to lenghten causing a restrinction in the free movement of it. The eyes usually look normal except in size gaze positions, when one eye appears higher than the other, particularly when looking up. It can be congenital or begin later. It can also be constant or intermittent.

Esotropia	84–90%			
		Alternating 70%		Monocular 30%
		Constant 57%	Intermittent 23%	Strabismus eliminated with glasses 19%
Exotropia	8–10%			
Vertical deviations	2–8%			

Figure 5.8: The frequency of the different types of strabismus when is present in Down syndrome (T. Watt et al.; 2014).

- **Is there any relationship between accommodation and strabismus?**

M. Cregg (1999) found that from the 38% of the subjects, who had strabismus, 24.2% had significant myopia (greater than -0,75D), 51.7% had significant hyperopia (greater than +2,75D) and 24.1% had no significant ametropy.
Therefore, the excessive accommodation to compensate for hyperopia, which leads to a development of strabismus hypothesis, does not explain the prevalence of myopes and those with non significant refractive errors in the strabismus group.
For this reason, the relationship between development of strabismus and accommodation needs further investigation.

In addition, when the visual system is less sensitive to blur, vergence-accommodation is the dominant factor in the accommodation system. While low spatial frequencies may be sufficient to drive the initial accommodation response, sensitivity to the high spatial frequency will be lacking and convergence-accommodation will therefore be responsible for making the response clearer. This imbalance in the vergence-accommodation relationship may play a significant role in the development of a squint.

5.4.2 Nystagmus:

Nystagmus was first described in Down's syndrome by Sutherland (1989) (N.R. Bromham et al; 2002).
Several investigators have found increased occurrence of Nystagmus in Down's syndrome, ranging from 5-30% (L. Averbuch-Heller et al; 1999).

5.4.2.1 Reasons of nystagmus development:

Although the cause of nystagmus in Down's syndrome is unclear, Pires da Cunha R. et al.; (1996) suggested in their studies that heart defects are related in some way to ocular manifestation of Down's syndrome, however no association was found between heart defects and accommodative insufficiency, hyperopia or strabismus. Myopia and

nystagmus can occur due to damage to the visual pathways. The study showed that children with Down syndrome with cardiac defects are more likely to have nystagmus than those without cardiac problems. In N.R. Bromham et al.; (2002) research, 9 out of 11 subjects who had nystagmus had heart defect. The myopic children were also more likely to have nystagmus than non-myopic, and 5 out of 6 myopes had nystagmus.

5.4.2.2 Literature review of nystagmus in subjects with Down syndrome:

Fierson WM et al. (1990) exerted ophthalmoscopy in children with Down syndrome and did not show any abnormality beyond the spoke-like blood vessel arrangement around the optic disc often seen in Down syndrome.

Nystagmus is reported in 8% (Olav H. H.; 2001) and 14% in Hiles and 3.3% for Woodhouse in subjects with Down's syndrome. Don C. Van Dyke et al.; (1990) study, nystagmus was present in 10% of the subjects.
In this same study, Jerk-type[2] nystagmus was the most common type of nystagmus in children with Down's syndrome and only one subject was reported to have rotary nystagmus[3]. No one had pendular nystagmus. And none of pendular cases of nysgamus was associated with optic nerve hypoplasia, which usually goes hand-in-hand with pendular nystagmus[4].
Also, L. Averbuch-Heller et al.; (1999*)* found that 23% of unselected adult subjects with Down's syndrome had Nystagmus and all cases were Latent/Manifest-latent Nystagmus (LMLN), which is far above the expected prevalence of LMLN in the general population. This type of nystagmus may reflect abnormal processing of visuospatial information, consistent with recent findings in patients and animal models of trisomy 21 (Randall T. J.; 1988).

In children with nystagmus, sometimes there is a position of gaze where the movements are considerably reduced. If this is the case, the child might adopt a compensatory position of the head in which the eyes minimize the movements, causing a possible torticollis. This can also be seen in children with Down's syndrome.
When there is a nystagmus affecting binocular vision, often it gets better in near than distance, that is why they may often prefer to hold books very close as this improves their vision and even if it might seem strange, they should be allowed to do this.
(Down's syndrome association medical series. Nº eye problems in children with Down's syndrome).

[2] Jerk-type nystagmus is a rhythmic eye oscillation characterized by a slow drift of the eyes in one direction that is repeatedly corrected by fast movements in the reverse direction.
[3] Rotary nystagmus: a slight movement of the eyes around the visual axis.
[4] Pendular nystagmus: type of nystagmus that in most position of gaze has oscillations of eual speedant amplitude, usually arising from a visual disturbance.

5.5 OCULAR PATHOLOGYES

5.5.1 Brushfield spots:

Are focal areas of iris stromal connective tissue hyperplasia surrounded by relative hypoplasia. They appear as speckled spots and are found up to 52% of the children with Down syndrome, although some studies claim that they are not present (T. Watt et al.; 2014).

They have no functional significance.

It is most common in those with light iridise and may become less visible with age if iris colour turns from blue to brown.

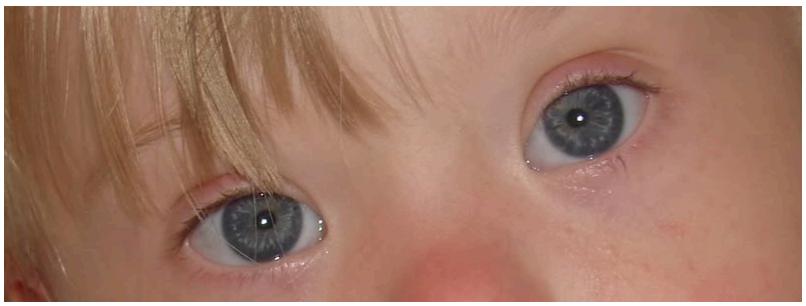


Figure 5.9: Brushfield spots in both eyes of a child with Down syndrome.

5.5.2 Blepharitis:

The presence of blepharitis in Down syndrome also seems to be common, with an incidence of 7-46%.

Creavin A et al.; (2009) reviewed some studies with Down syndrome, which provided data for the prevalence of blepharitis. This study showed that in 6 out of 11 subjects the prevalence was 10% or less, in 3 out of 11 was 15-20% and in 2 out of 11 was 15-30%.

Figure 5.10: Eyelashes with blepharitis.

5.5.3 Cataracts:

Although some studies may demonstrate a low prevalence of cataracts in early age in Down syndrome children, it is still more common than in normal subjects. Furthermore, congenital cataracts are described to be common in DS subjects (B Haargaad et al; 2006, T. Watt et al.; 2014).

5.5.3.1 Literature review of cataracts in subjects with Down syndrome:

The estimated frequency of early cataract in Down syndrome is 1.4% (B Haargaad et al; 2006). B. Haargaad et al.; (2006) supported that early cataract is a rare event in Down syndrome, though 1/3 of 29 children with Down syndrome of his research had their cataract diagnosed already at birth. The 2.8% of these cataracts were non-traumatic and non-acquired cataract. By contrast, Roizen NJ et al.; (1994), Igersheimer J.; (1951), Pearce FH et al. (1996) and Berk AT et al. (1996) found that the occurrence of early cataract among children aged up to 17 years with Down syndrome was reported to be of 5% to 50%. To give more evidence, a *Danish* study demonstrated a prevalence of 1.4% of cataracts in Down's syndrome at birth, compared to general population to be 0.06% (T. Watt et al.; 2014).

In previous studies of congenital or infantile cataract, 3-5% of cases were associated with Down syndrome (Bhatti TR et al; 2003, Merin S et al; 1971, Wirth MG et al; 2002, Rahi JS et al; 2000). However, there is a lack of population-based data on occurrence and characteristic of early cataracts in Down syndrome (B. Haargaad et al; 2006). In a recent UK study of congenital and infantile cataract, 5.4% were Down syndrome patients in which 61.5% of them were diagnosed with cataract in the neonatal period (Rahi JS et al; 2000).

Nuclear cataract does not cause a significant problem and is relatively common in people with Down's syndrome. A denser opacity of most of the centre of the lens is fortunately less common as it causes a marked reduction in vision. Less than 1% of children with Down's syndrome have a dense cataract.

5.5.3.2 Management:

Not all of the cataracts in babies and children need to be removed. Some cataracts are small and or off-centre in the lens. When the cataract is left in place, the vision still develops normally with no need to remove it. However, if vision seems to be affected, it should be removed as soon as possible, as it can interfere with normal development of the vision centres in the brain. Younger children may require an additional opening in the posterior lens capsule with some vitreous gel removal. An intraocular lens is then sometimes placed within the empty lens capsule.
If a lens implant is not inserted, the eye needs to be focused wither by wearing thick glasses or contact lenses (Down's syndrome association Medical Series. "Eye problems in children with Down's syndrome. Notes for parents & carers"; 2007).

5.5.4 Keratoconus:

Rados was the person who first reported the association of keratoconus and Down's syndrome in the ophthalmic literature in 1948 (M. Madison Slusher et al.; 1968). This condition is recognized to be more common in children with Down syndrome than in general population (M. M. Slusher et al; 1968), although it still relatively rare.

During the early stages, this condition makes the person short-sighted, often with marked astigmatism. Many cases do not progress any further than this stage. On the other hand, other cases go on to develop scarring in the centre of the cornea (J. M. Woodhouse et al.; 1997).

The condition is extremely rare in childhood, for example, in J. M. Woodhouse et al.; (1997) study no children with DS appeared to have keratoconus. However, this may be explained because of the late developing, as it may start to develop in adolescence, and ultimately affect 10-15% of adults.

Although it is rare, it is very important for subjects with Down syndrome to have regular eye checks throughout the teenage years and beyond.

5.6 COLOUR VISION

It is seen that Down syndrome subjects show a substantially higher incidence of colour vision deficiency compared to subjects with similar mental retardation.

A.J. Adams et al.; (1993) used the Ishiara plates and the Davico's anomaloscope[5] to test colour vision in Down's syndrome population. Their findings showed a significantly high portion of colour deficiencies in Down's syndrome group compared to normal children or matched population.

The research found that only 79% of the subjects with Down syndrome were able to perform Ishiara and only 67% could be tested with the anomaloscope. From those who could have colour vision tested, 23% had defective colour vision. Pérez-Carpinell J. (1994) found an incidence of 18% for colour vision deficiency. However other studies, such as Lowe; (1949), Stratford and Mills; (1984) differ in the findings of the high incidence of colour defects. Nonetheless, in A.J. Adams et al.; (1993) among those who were found a colour vision deficit, 10/13 were protan, 1/13 was deuteranomalous and 2/13 had no deficit.

[5]Davico's anomaloscope is a recently developed system which uses light emotting diodes as the light source and electronic control over the luminance of yellow and the proportion of red/green to perfofm a Rayleight-type match and is used to detect color vision abnormalities in deutans or protans.

6 CLINICAL ASSESSMENT

As explained in 4.1.3 Down syndrome subjects are known to suffer from ocular and vision problems among other health problems.

It is crucial to carry out a visual examination as soon as possible in order to determine if there is any ocular anomaly, and give the best refractive correction or management and avoid potential problems in the development of these children. Routine vision assessments should start from birth to confirm the type of Down syndrome, a chromosomal study "in situ" to determine the type of trysonomi and make a family genetical study. Visual check should start at the age of 6-12 months and repeat every year until the age of 6 years of age (CSDC; 2005, Federación española del syndrome de Down (FEISD), J. Puig et al.; 2002)(table 6.1) and thereafter, every two years.

The Health Program for persons with Down syndrome from Catalonia (Programa de salut per a persones amb syndrome de Down) and the Down's syndrome Medical Interest Group (DSMIG) (CSDC; 2005, Down's Syndrome Association Medical Series; 2007, Guía oftalmológica del syndrome de Down, Dr. José Mª Borrel Martínez) recommends to follow this eye check routine in Down's syndrome children:

	Birth and 6 weeks	6 weeks-12 months	12 months	18-30 months	3-3.5 years	4-4.5 years
Eye check	Visual behaviour. Check for congenital cataract.	Visual behaviour. Check for squint.	Visual behaviour. Check for squint.	Orthopic examination. Refraction and ophthalmic examination.		Visual acuity, refraction and ophthalmic examination.

Table 6.1. Table of eye check recommendation from DSMIG (Down's Syndrome Association Medical Series; 2007).

Some of the difficulties the optometrist may face when testing Down syndrome children are communication and their limited capability to pay attention. Also, when the task becomes more complicated, they try to avoid making a mistake, which means that the results may not be exactly what they can reach. So when examining, optometrists should take into account that the eye test must be adapted to objective methods, tests adapted to their cognitive level, the need to use other abilities to catch the attention of the child, to be flexible and patient during the exam, to present the tests like a game and to encourage to keep trying to do their best. Optometrists have to be dynamic and quick when examining and get the results as fast as possible, in order not to tire the patient.

Tests should be done objectively as the response given by Down syndrome children may not be trusted, as we can not know whether the child is able to see the target or unable to keep doing the tasked asked, or whether he is just tired and does not want to keep trying when the task gets more difficult.

It is important to use the appropriate test for the subject's age and collaboration. It is necessary to cheer the child and encourage to keep on trying harder when the task gets more complicated. This will possibly give better results of his visual acuity.

Not all exams can be done in Down syndrome subjects, as more time would be required and our aim is not to spend a lot of time doing the eye test, because the more tired the child gets, the worst the results would be. The tests for an eye exam for Down's syndrome should include: history & symptoms, visual acuity, retinoscopy, binocularity exam, accommodation examination, stereopsis and ocular health.

6.1 HISTORY & SYMPTOMS

It is important to do a good and complete history case and symptoms, because it is where the professional can determine the matter of the visit and know about ocular, medical, personal and family history. It is also a first opportunity for the optometrist to interact with the patient and gain his or her trust.

It will be difficult to obtain direct information from Down's syndrome patients about their complaints, so the optometrist should also ask for information to the parents or the caregivers. This means that the information that we can possible get it is based on the observations of others. Some questions can be asked to the child if he collaborates.

6.2 VISUAL ACUITY

When testing visual acuity in Down's syndrome cognitive ability, age and cooperation must be taken into account. Tests have to be adapted to their ability in order to make the exercise easier for them.

Some of the tests that can be used in these subjects are: HOTV matching, forced choice which using forced choice or matching strategies is useful in preschool children, Kay Picture Test, Crowded LogMar Book, Broken wheels from Richman.

Some of them may need a little familiarization with it, so it would be good to try a couple of times to prove the child understands what he has to do or just to identify the figures and get familiar with them.

Something we should be concerned about in Down syndrome children is that even with fully corrected refractive errors and in the absence of manifest abnormalities, they have reduced acuity compared to their developmentally healthy peers (J.A. Little et al.; 2007).

6.2.1 HOTV matching:

HOTV matching test is a visual acuity test designed by Sheridan-Gardner which is an excellent test for children who are unable to perform vision testing verbally. It is based on a Snellen card, with letters that have no-directional, which makes it easier to identify. The advantage of this test is that it is not verbal.

Distance: from 3 to 6 meters
Mono/Binocular: monocular and binocular
Conditions: good light, with the habitual correction
Material: occluder, HOTV test

Figure 6.1: HOTV matching test.

6.2.2 LEA symbols folding chart:

This is a visual acuity test for distance vision held at three meters distance. This can be especially useful in children between 3 to 5 years of age or with subjects who are not familiar with the alphabet.

The test is based on four symbols (apple, square, circle and a house), which gets blurred equally at the threshold. When the subject no longer can recognize correctly the symbol, it transforms into circles or “rings”. The symbols are easy to name, point or sign.

It is used in Down syndrome children as there is no need in knowing the letters and it gives comfortability and sense of playing a game.

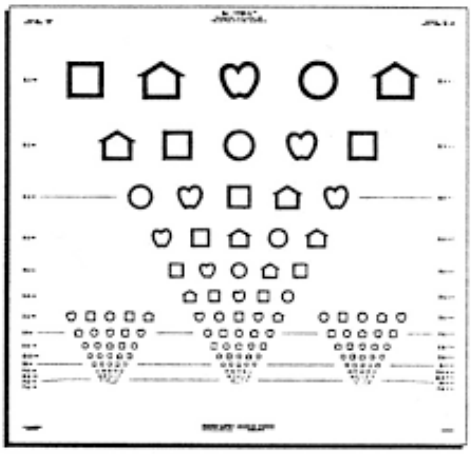


Figure 6.2: LEA symbol test, 15-folding chart. (Vistest (SF-022200 Espo Findland)).

6.3 REFRACTIVE ERROR ASSESSMENT

The assessment of refractive status in very young children is often not conducted in the same manner as for adult patients. In particular, the child's age, their cooperation and dynamic refractive status will be key factors, which influence the accuracy of refraction. For this reason, it is often necessary to choose procedures, which inhibit or minimise accommodative activity. The same happens between subjects with cognitive disabilities and any disability, for example, Down's syndrome will not have the same collaboration than normal subjects.

This exam is of utmost importance because, as referred in chapter 6, there is a high prevalence of refractive errors in subjects with Down syndrome compared to normal subjects. It can be seen early in preschool age. Thus, in order to give the refractive correction as soon as possible and avoid a delay in development of these children, an examination of the refractive error must be carried out. This is why refractive error assessment should be performed since they are infants (table 6.1).

The most appropriate way to check for refractive error is Mohindra retinoscopy, which will be explained lately, and wet retinoscopy with cyclopaedic agent. It is an objective exam, which allows the optometrist to obtain information of the refractive error of the person.

Mohindra retinoscopy is the most used technique for retinoscopy in Down syndrome subjects, especially in children, as there is no need to use cyclopaedic drops into the eyes of the children avoiding the secondary symptoms and the uncomfortable sensation.

In this part will give more emphasis in Mohindra retinoscopy technique because this is the one that will be used in the experimental procedure:

6.3.1 Mohindra retinoscopy:

The Mohindra retinoscopy, also known as near retinoscopy or near monocular retinoscopy, is a technique used in young children to measure the refractive error. It is much more child-friendly and requiring less co-operation from the child (Fabrizio Bonci et al.; 2012).

In this case, the stimulus is the dimed light source of the retinoscope in a darkened room. The darkness of the room will facilitate the child to keep looking on the retinoscope light. This test has to be done at 50 cm of distance. A skiascopy will help with the dinamisation of the test as there is no lost time with changing lenses and the child gets less tired. As the Mohindra technique relies on the observation in total darkness, the eyes establish a small amount of accommodation and equally in both eyes.

As Mohindra takes place in total darkness, a few children might find it scary, but with appropriate preparation and explanation this will rarely be a problem. So begin explaining what is going to happen and that lights will turn off. If a parent is close this will give trust to the child. It is recommended to lower the lights gradually keeping the retinoscope in the eyes of the child all the time.
The technique should be performed monocular, although Wesson MD et al.; (1990) demonstrated that there is no difference in the result if binocular fixation is allowed. Trial frames can be used or a simple patch.

It is important that during the examination the light of the retinoscope is kept on the child's pupil, so that accommodation is not stimulated (Fabrizio Bonci et al; 2012). Maximum size of the pupil will indicate no accommodation. Afterwards, the aim is to neutralize the reflex seen through the retinoscope.
Trial frames sometimes can be uncomfortable to Down syndomre children due to the physognomy of their nose.

When calculating the value of the refractive error (+1.25D) has to be taken away from the gross finding (Fabrizio Bonci et al; 2012). This value that has to be discounted was determined by Indra Mohindra, who calculated empirically in clinical studies while doing this technique.

In a normal child we would expect to find a refractive erorr of +0.50D while in Down syndrome would be higher, due to the failure of emmetropization.

Distance: 50 cm
Mono/Binocular: monocular
Conditions: completely dark room
Material: retinoscope
Other tests: cyclopaedic retinoscopy

6.4 ACCOMMODATION

Accommodation is reported to be deficient in Down syndrome subjects and most of them under accommodate (5.3). Dynamic retinoscopy is a good exam to determinate the subjects near point of accommodation.

6.4.1 Dynamic retinoscopy:

Dynamic retinoscopy is a Nott test modification, which determines the patient's near point. In other words, it looks for the location in space that a patient's eyes are focused when fixating a near target. It is primarily used to confirm suspected cases of vergence and/or accommodative dysfunction. It also reveals the stability or the degree of fluctuation of the accommodative spasm.

When the accommodation is hanging out in front of the target, the accommodative response is bigger than the stimulus (accommodative lead), while when the accommodation is hanging out behind the target then the accommodative response is smaller than the stimulus (accommodative lag). Lag of accommodation is very common in children with Down's syndrome, where it is expected to find neutral move behind the stimulus.

In normal children it is expected to find a value of +0.5D. It can vary depending on the test used, for example, in MEM a normal value of +0.75D is expected to be found in normal children and in cross-cylinder +0.5D. However, in Down's syndrome this value is expected to be higher, around +1.00D or greater.

Distance: around 40 cm, but it varies with the test
Mono/Binocular: binocular
Conditions: refractive error correction in place
Material: retinoscope, target (fixation target/visual acuity test adapted to the person)
Other techniques: Nott, MEM, cross-cylinder

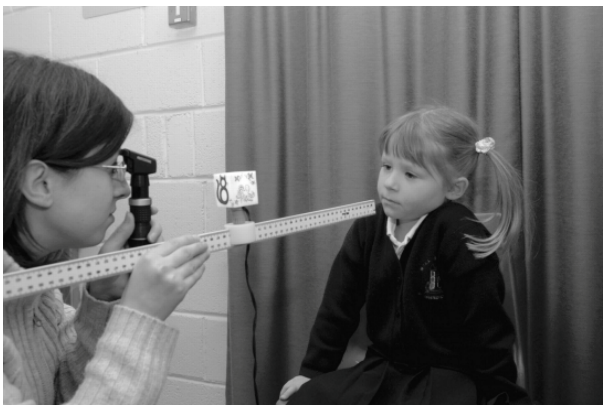


Figure 6.3: Dynamic retinoscopy in a child.

Distance of target from patient	Distance of the retinoscope from the patient when a neutral reflex is observed
40 cm (AS=2.50 D)	50 cm (AR=2.00 D)
25 cm (AS=4.00 D)	33 cm (AR=3.00 D)
20 cm (AS=5.00 D)	25 cm (AR=4.00 D)
17 cm (AS=6.00 D)	20 cm (AR=5.00 D)
14 cm (AS=7.00 D)	20 cm (AR=5.00 D)
13.5 cm (AS=8.00 D)	20 cm (AR=5.00 D)

Table 6.2: Assessment of the amplitude of accommodation using dynamic retinoscopy. For each target distance, the accommodative response (AR) is determined by finding the position of the conjugate with the retina. Eventually, a point will be reached where further increases in the accommodative stimulus (AS) are not accompanied by an increase in the AR (M. Rosenfield et al.; 2009).

6.5 BINOCULAR VISION ASSESSMENT

Binocular vision is really important to be evaluated in Down syndrome subjects because, as said in chapter 5.5.1, they have a higher prevalence of squints in comparison to normal subjects.

Relevant tests to assess Binocular Vision in Down's syndrome include: Cover Test (CT), Near Point of Convergence (NPC) and Ocular Motility Balance (OMS) and stereopsis.

6.5.1 Cover test (CT):

It is an objective test aimed at detecting and measuring the presence of strabismus or phorias. It uses an opaque occlude to break fusion.

In this test, a fixation target is needed to draw the attention of the child and also to stimulate accommodation. This target can be an optotype adapted to the age of the person and his visual acuity. In children it can be a small toy with some details in it.

The ideal thing is to perform cover test, both at far and near distances, as the results can be different for each distance, due to the accommodation. Also it is necessary to examine at different gaze positions to check for incommitancy.

Distance: far, intermediate or near
Mono/Binocular: binocular
Conditions: good light
Material: occluder, fixation target to activate accommodation
Other techniques: cover/uncover, CT alternate, unilateral covert test

When there is no binocular vision and there is a squint instead, it can be measured with cover test, Hirshberg or Krisky tests.

6.5.2 Near Point of Convergence (NPC):

The Near Point of Convergence is an objective or subjective test, which measures the ability to converge while keeping fusion on a fixating target which is moved closer to the subject. It is frequent to find some people that still can see single when the object has reached the nose.
The standard value for NPC in normal children ranges between 5 to 10cm/ 15-10cm and increases 0.24cm/year until fifteen years old.

The point of fixation can be a light, a red filter, a little object or even Snellen letters.

It is measured in near vision, starting from a distance of 40cm and moving the target closer until the break up or the nose if it is the case.

In Down syndrome children we have to be aware that we might no get a response from the child, so we have to be careful and observe the eyes of the child and note if there is any deviation in one of the eyes and when it happens.

Distance: from 40 cm getting closer
Mono/Binocular: binocular
Conditions: good light
Material: object to observe

6.5.3 Ocular Motility Balance (OMB)

It is a test with the aim to find any restriction in the muscles of the eye that can cause an abnormal eye position.

In this exam the patient has to follow (with the eyes and without moving the head) the light that is moving from side to side smoothly.
If there is any slowness or inaccuracy in his ability to follow the visual targets it has to be recorded. The eight positions of gaze have to be examined.

Distance: 40 cm
Mono/Binocular: binocular
Conditions: good light
Material: punctual light or object

6.6 STEREOPSIS

Stereopsis is checked in Down's syndrome to discard suppressions and check the integrity of the binocular function.

There are several useful stereotests to measure stereopsis in Down's syndrome. The most used and recommended is Frisby stereotest, although with adaptation to the capability of the subject all of them can be appropriate.

6.6.1 Frisby stereotest:

It is a useful test in children to discard suppressions and check for stereopsis.

It consists of three plates of different thickness, which present transparent stereograms of real depth. They are made with two different images with horizontal disparity.
The stereopsis acuity varies with distance and can reach 15 to 600 sec arc.
One disadvantage of this test is that it can introduce monocular clues. However, the advantage of this test is that no special glasses are needed so it makes it suitable for babies, children or children with special needs. It is also simple and easy to administer and can be done even when amblyopia is present.

In order to give motivation to the child, a sound can be used whenever the child guesses right. This is a modification of Frisby stereotest.

Distance: 30cm to 150cm
Mono/Binocular: binocular
Conditions: good light, correction for refractive error
Material: Frisby stereotest, sound stimuli
Other techniques: Random Dot E (RDE), Lang, Randot

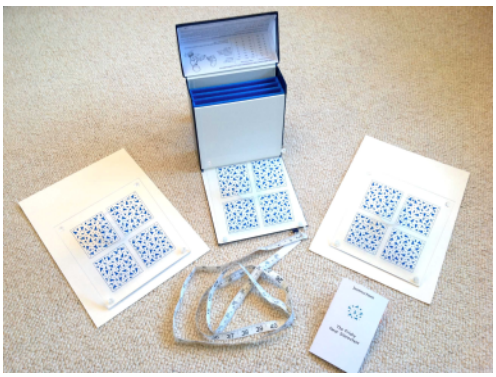


Figure 6.4: Frisby stereotest.

Figure 6.5: Down syndrome child being examined with Frisby stereotest

6.7 COLOUR VISION

Defective colour vision has also been reported to be present in Down's syndrome.

Lots of tests can be used when testing for colour vision, for example, Farworth-Munsen D-15, Ishihara plates and the anomaloscope. Obviously, many of them are useful to detect colour blindness and are adapted to Down's syndrome subjects. However, Ishihara Colour Vision Test is the most common, therefore as the test chosen for our assessment.

6.7.1 Ishihara Colour Vision Test:

It is the most well-known colour blindness test and the most widely used. It consists of a set of coloured plats, which are made of dots with each one showing a number or a path.

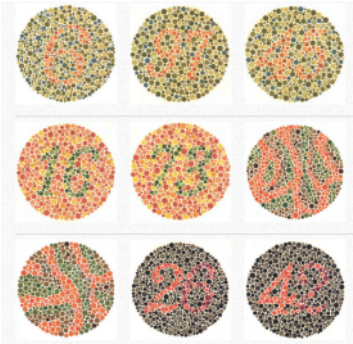


Figure 6.6: Some of the plates shown in Ishihara's colour vision test.

6.8 OCULAR HEALTH

As Down syndrome subjects are known to suffer from blepharitis, brushfield spots, cataracts and keratoconus, among others ocular health problems, it is recommended to check for ocular anomalies.

Pupil reflex, observation of the ocular surface with slit lamp to check the integrity of the cornea, the presence of blepharitis in the eyelashes and the presence of cataracts and keratoconus are some important condition to look for in an ocular health examination as well as to asses the state of eyelashes and eyelids and ocular fundus. In case of refraction under cyclopaedic drops, ophthalmoscopy can also be used to examine the retina.

7 METHODOLOGY

This small study focused in detecting and analysing the vision deficiencies found in a population of Down syndrome.

My short stay at the University of Optometry of Cardiff during, allowed me to participate with Down syndrome and mentally disabled subjects with professionals such as J.M. Woodhouse and the PhD working with here, in order to get the necessary knowledge and skills to be able to examine adequately a subject with Down syndrome.

This part aims to explain how clinical assessment was practised to the 22 subjects with Down syndrome from Fundació Down de Lleida and which techniques were used.

It also gives a brief mention to the ethical principles in the accomplishment of experimental studies of this study and a brief introduction about the small sample examined.

7.1 ETHICAL PRINCIPLES IN THE ACCOMPLISHMENT OF EXPERIMENTAL STUDIES

Before performing any type of investigation, it is necessary to know the ethical, legal and juridical requirements when the investigation is done in humans. The “Associació Mèdica Mundial” (AMM) has enacted the Declaration of Helsinki as a proposal of ethical values for the medical investigation in humans, including the investigation of human material and identifiable information. This is why this study will be based in the Declaration of Helsinki, a very important international document in the ethical biomedical investigation applicable since June of 1964 what is used when several studies are realized with humans. (Associación Médica Mundial (AMM), 2008).

The Llei Orgànica 15/1999, of 13en of December, of data protection and the law 41/2002 from 14en of November, regulatory of the autonomy o the patient and his rights and obligations in the information and clinical documentation which containing the guidelines to be followed to implement the duty secret were used when protecting the personal data of the subjects participating in the study.

The tests done are not invasive, nevertheless they were all explained to the subjects or their legal representatives and everyone of them read the information sheet and signed the informed consent.

7.2 SELECTION OF SAMPLE

The subjects of the sample investigated in this study were Down syndrome subjects attending Fundació Down Lleida, which is a day centre for Down syndrome subjects. The age range was between 6 to 40 years of age and the final sample had 22 subjects.

Some of them are actually living in foster homes with a high level of independence and understanding.

7.3 EXPERIMENTAL PROCEDURE

All subjects underwent a throughout visual exam in order to determine the presence of refractive errors (myopia, hyperopia and astigmatism) the accommodation response exerted at two different distances (20cm and 40cm), stereoacuity and colour vision discrimination. The techniques used are the following:

- Visual acuity
- Over refraction
- Dynamic retinoscopy
- Frisby stereotest
- Ishihara colour vision test

The examination was performed in a quiet, pleasant room of the Fundació Down Lleida Center. It was agreed with personal, parents and volunteers in the Center, that cooperation would increase if the study was performed in a familiar and comfortable environment.

7.3.1 Visual acuity measurement:

Visual acuity was measured with the Light House visual acuity chart Test (10-folding lines) at a 3m distance. This test consists of a wall chart which displays a square, a circle, an apple and a house as optotypes and it was presented at three meters distance from the subject (fig 6.2).

The subject was sitting in a chair while the examiner was pointing to a symbol on the wall chart and the subject had to identify and say which symbol was. If the child wasn't cooperative or seemed to have difficulty in saying the name of the figures or didn't want to talk, it was provided a matching card with the figures and was asked to point at the symbol matching the one with pointed on the testing board.

Visual acuity was measured monocular and binocularly. An occluder was given when testing visual acuity monocularly, making sure they occluded correctly. For those who had problems when occluding, they were told to do it with their hand.

Twenty out of 22 of the subjects were wearing spectacles full-time; the other two didn't wear glasses at any time. Those who wore spectacles had their visual acuity measured with correction and those two who without, did not.

When a subject couldn't identify any symbol from the first raw, equivalent to decimal visual acuity of 0.2, the viewing distance was changed moving the test towards the patient at a distance of 1.5m, and the visual acuity was checked again and noted.

7.3.2 Over refraction:

To make sure all subjects were wearing the correct and full correction, over refraction technique was performed over their glasses. The examiner was situated at a 40cm distance from the subject and a +2.00D lens was situated in front of one eye to blur the image and block the use of accommodation. Then refraction was performed in the other eye.

While doing over refraction, the subject was observing a target of 0.2 decimal visual acuity target of the LEA-visual acuity chart (fig 6.2).

When a subject was found an over refraction value greater than 1D, the new refractive error correction was put in the trial frames and used for the rest of the assessment, as we were looking for the best optical correction to be used for the rest of the assessment.

7.3.3 Dynamic retinoscopy (Nott):

Dynamic retinoscopy is an objective technique to estimate the accommodation response accuracy and amplitude to a near target. It consists of presenting a fixation target that stimulates accommodation at a distance of 40cm (2.50D) and to move towards or forward to the patient until finding neutral, which indicates where accommodation response to the target is. All patients have to wear the full correction in their specs to avoid getting wrong and not trustable results.

If the accommodative response is accurate (i.e. accommodative response exactly on the accommodative stimulus), then the reflex of the retinoscopy in the eye will be neutral. However, if there is under-accommodation a "with" movement will be seen. In this case, it is necessary to move further from the subject until neutrality is found. If the subject over-accommodates, an "against" movement will be seen and it will be necessary to move closer to find neutral reflex. The difference between neutral and the target will represent the lead of accommodation.

In this case, the exam was done at two different distances, 20cm (+5D) and 40cm (+2.5D). Some studies have shown that Down syndrome individuals have a mean lag of accommodation greater than +1D between distances of 20-30cm, so the aim was to determine the accommodative response for both distances to investigate the difference in accommodative response.

All subjects were sitting in the chair and the examiner was in front of them. All were wearing their current specs correction. The individuals were presented a near vision

target at 40cm first and later at 20cm. The target was a fixation stick with little detailed drawings. The subjects were asked to identify one of them while dynamic retinoscopy was done. The same procedure was repeated for the other eye and for the second distance. The distance at which neutral was found was noted.

7.3.4 Frisby stereotest:

It is a simple and effective screener and assessment test of stereopsis vision, which determines stereoacuity and helps to discard suppressions. Moreover, there is no need for special glasses while performing the test and it is suitable to use on young children, and mentally disabled subjects, as are Down's syndrome.

In order to examine stereopsis, the examiner explained to the individual what was the test about and what he could expect with an example to allow familiarisation with it. Afterwards, the subject was shown the first plate (6mm) at 40cm distance and was asked to point in which of the squares there was a circle coming out. If they could answer correctly the exam proceeded with the thinner plate (3mm), however if the subject was not able to see the first plate, this one was moved closer towards the patient at a distance of 30cm and was asked to do the same question.

7.3.5 Ishihara 38 plates Colour Vision Test:

In order to detect colour vision anomalies in our sample, the Ishihara (38 plates) colour vision test was used (fig 6.6) for this assessment. The test was presented at a distance of 40cm. The subjects were asked to identify the number in the several charts presented. If they were unable to say the number, they were asked to follow it with the finger and in extreme cases the paths plates were presented. If no numbers were identified or the number was incorrect, it was recorded as a failure.

8 RESULTS

In this study, 22 subjects were examined, 12 of whom were female and 10 were male. The age of the sample varied from 8 to 40 years of age, which was distributed as follows:

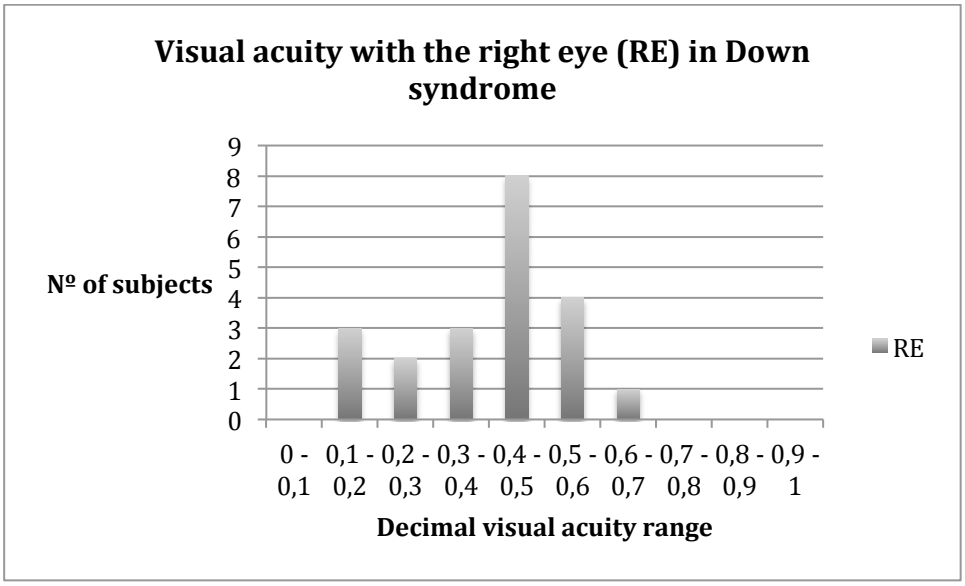
GROUP BY AGE:	Nº OF SUBJECTS	FEMALE	MALE
Children (5 to 12 yr)	2	0	2
Adolescents (13 to 18 yr)	5	3	2
Young adults (19 to 29 yr)	10	4	6
Older adults (30 to 40 yr)	5	3	2

Table 8.1. Distribution of Down syndrome subjects examined in Fundació Down de Lleida Centre into groups by age.

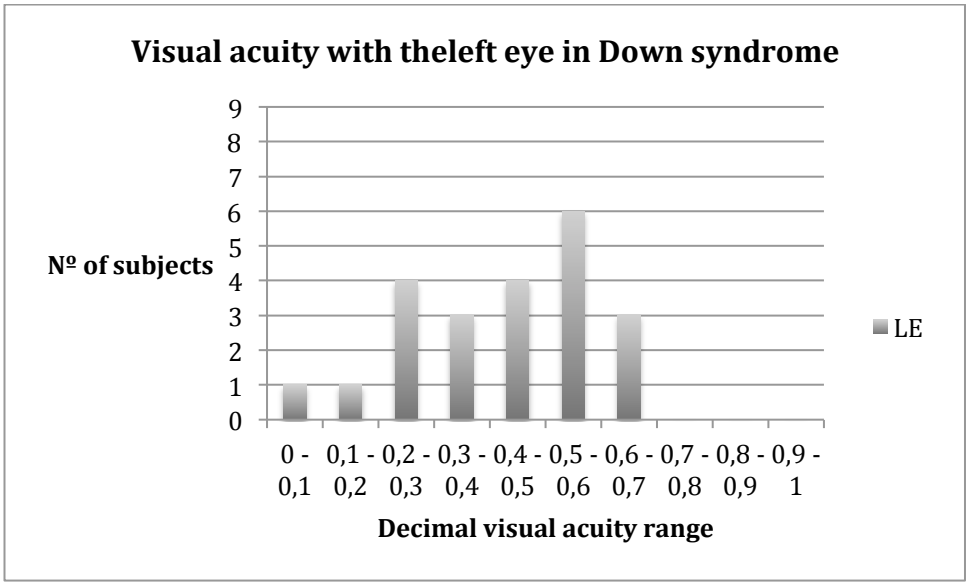
Twenty out of 22 subjects already wore a refractive error correction.

8.1 VISUAL ACUITY (VA)

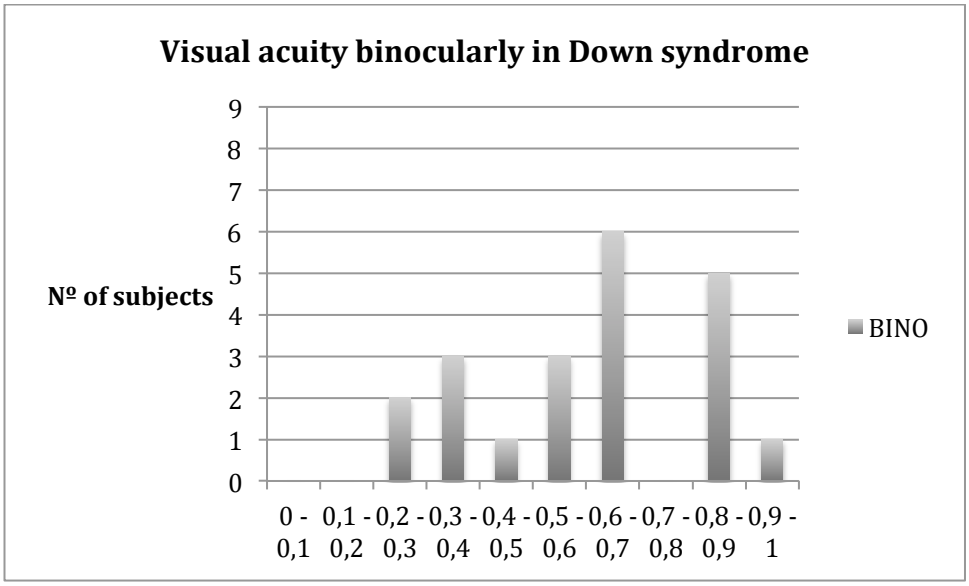
In this part shows the results for visual acuity. Graphics 8.2a, 8.2b and 8.2c show the frequency for VA decimal values monocularly and binocularly.



Graphic 8.1a: Frequency of corrected decimal VA for right eye (RE) in a group of DS subjects.



Graphic 8.1b: Frequency of corrected decimal VA for left eye (LE) in a group of DS subjects.



Graphic 8.1c: Frequency of corrected decimal VA for both eyes in a group of DS subjects.

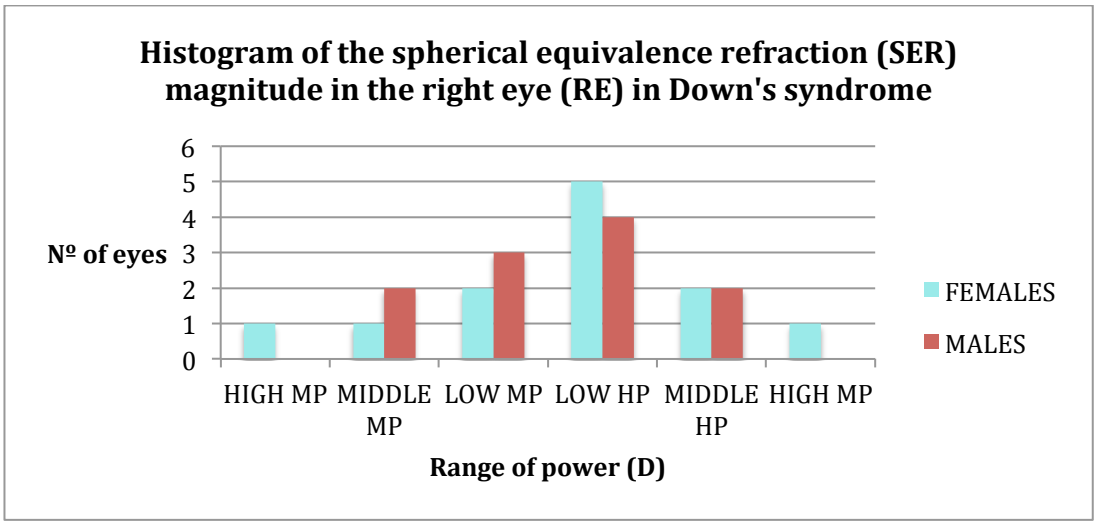
As it can be appreciated in graphics 8.1a, 8.1b, 8.1c, in the right eye the more frequent value for visual acuity is 0.4-0.5 in the decimal scale, followed by values of 0.5-0.6. In less proportion but also common are values of 0.1-0.2, 0.3-0.4. Only one subject overpassed the value of visual acuity of 0.6 (0.63). In the left eye the values obtained for visual acuity differ from the right eye. In this case we see that there is not a more frequent value and the results spread over different values. For example, although the more prevalent visual acuity is 0.5-0.6, we see that there are also several cases with visual acuities of 0.2-0.3 and 0.4-0.5.

The mean values for visual acuity were 0.29($\pm$0.10), 0.29($\pm$0.12)monocularly and 0.50($\pm$0.17) binocularly.

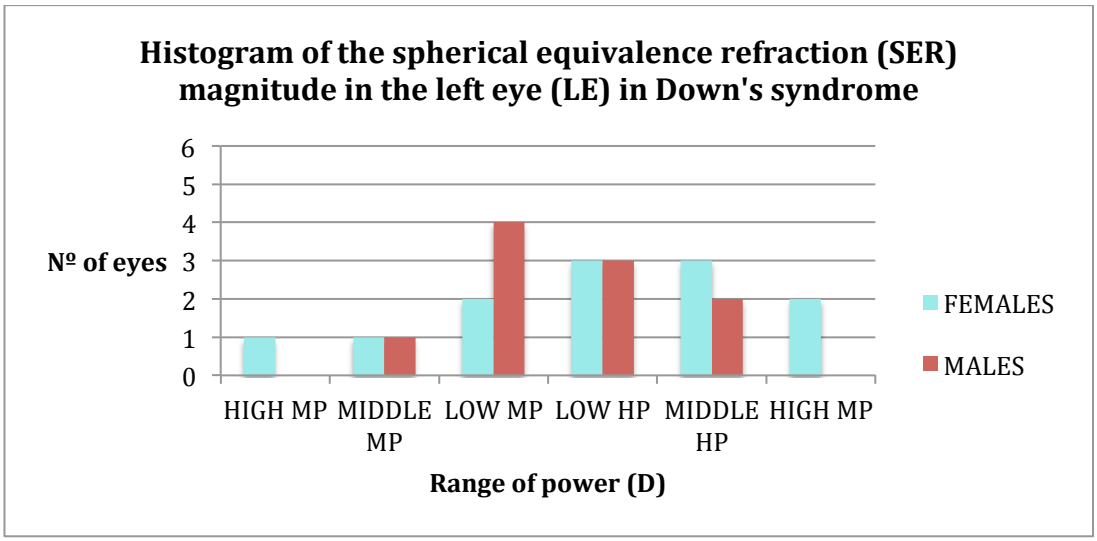
8.2 SPHERICAL EQUIVALENT REFRACTION (SER) AND ASTIGMATISM

Graphs 8.2a. and 8.2b. show the distribution of Spherical equivalent refraction (SER) in right and left eye according to sex for all subjects examined.

Hyperopia and myopia has been classified into three groups: low, middle and high spherical equivalent refractive errors (SER). Low hyperopia ranged from 0 to +2D, middle hyperopia from +2.25D to +5D and high hyperopia for values higher than +5D. The same for myopia, being low for values between 0 to -3D, middle from -3.25D to -6D and high myopia for values higher than -6D.



Graphic 8.2a: Frequency of refractive error in right eye (RE) for DS group of subjects studied.



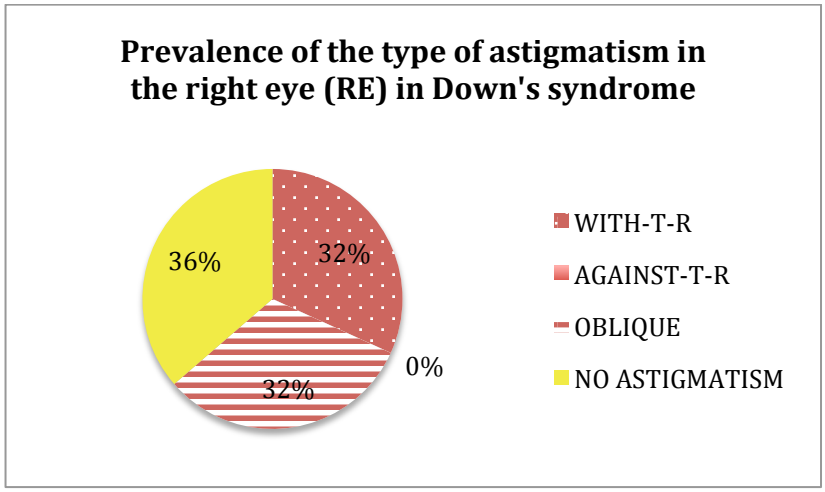
Graphic 8.2b: Frequency of refractive error in left eye (LE) for DS group of subjects studied.

Graphic 8.2a and 8.2b show the frequency of refractive errors (SER) in the group of Down syndrome subjects studied for right and left eye.

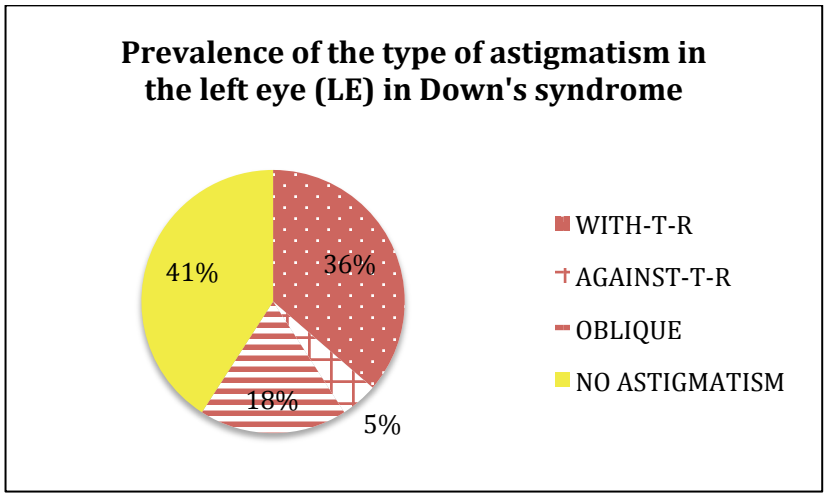
As it can be appreciated, the most frequent refractive errors are between -3.00D and +2.00D in both eyes. Although the frequency of hyperopia is higher in female subjects for the right eye, male subjects have a higher frequency of myopia refractive errors (SER). For the left eye, there is no difference for values of 0 to +2.00D between men and women, however the frequency of myopia (SER) between 0 and -3.00D is higher in male subjects than in women subjects. Another aspect to point out is that only few subjects had refractive error (SER) greater than +5.00D or -6.00D, and they were only female subjects.

The mean hyperopic refractive error (SER) in the right eye is about $+2.39 \pm 1.65$D and $+2879 \pm 1.52$D in the left eye, while the mean myopic refractive error (SER) is -39 ± 1.85D and -2.709 ± 1.69D respectively.

Astigmatism was also found in these subjects. Astigmatism was considered to be present for values greater or equal to ≥ 0.75DC. The frequency and type of astigmatism was investigated.



Graphic 8.3a: Frequency and type of astigmatism present in the right eye (RE) of the DS subjects studied.



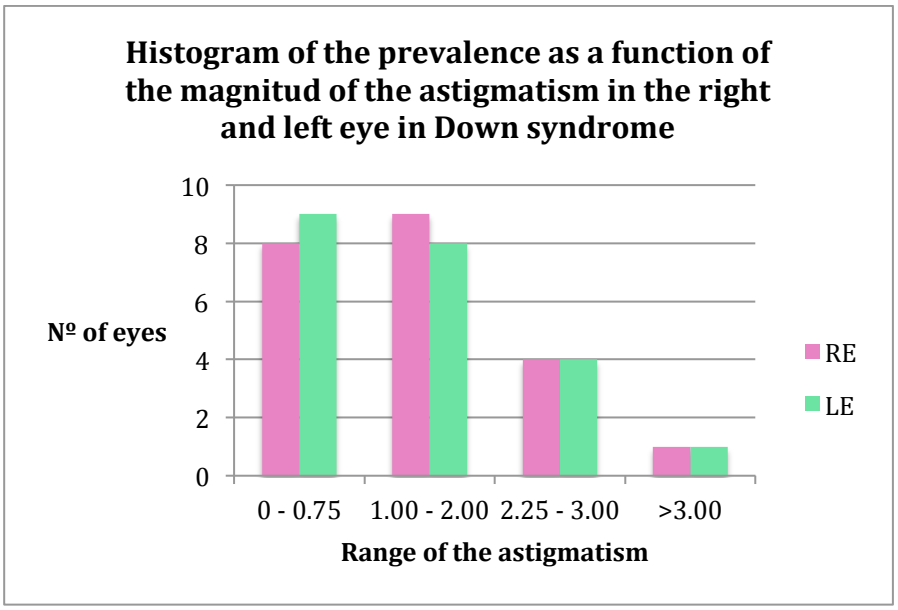
Graphic 8.3b: Frequency and type of astigmatism present in the left eye (LE) of the DS subjects studied.

Graphic 8.3a and 8.3b show the percentage and type of astigmatism in the population of Down syndrome studied, for the right eye and the left eye.

As it can be seen, the percentage of subjects with low astigmatism (or no astigmatism (<0.75) is around 40% for right and left eye. Mean value for those subjects considered to have astigmatism is around 2.00DC.

The distribution of type of astigmatism is nearly 1/3 for with-the-rule, 1/3 for oblique and 1/3 of subjects had no astigmatism of <0.75DC. There was no case of against-the-rule astigmatism for the right eye. The left eye also has nearly 1/3 of with-the-rule, 28% of oblique astigmatism and a 5% of against-the-rule astigmatism. Nearly 1/3 of cases had no astigmatism of <0.75DC in left eye.
It has also been seen that, in the right eye, 4 out of the 14 eyes with astigmatism followed the pattern of the axis at 135º±10º caused by the shape of the eyelid, and 2 out of 13 left eyes presented oblique astigmatism (45º±10º).

Graphic 8.4 shows the frequency and distribution of the magnitude of the astigmatism found in the group of Down syndrome subjects examined. The grades for astigmatism are divided into: low astigmatism for less than 1DC, moderate between 1-2DC, severe between 2-3DC and extreme for values higher 3DC.



Graphic 8.4: Histogram of the frequency of the magnitude and distribution of the magnitude of the astigmatism, for the right and left eye, in the Down syndrome sample examined.

The highest frequency magnitude of the astigmatism for both right and left eye lies between 0-0.75DC and 1-2DC, thus low-moderate or no astigmatism are the most common types. Only one subject had astigmatism greater than 3DC in both eyes.

8.3 ACCOMMODATION RESPONSE

The quality and accuracy of the accommodative response was assessed with the Nott test (see section 7.3.3), which determines the lag of accommodation at a specific near distance. As mentioned in section 5.3, the lag of accommodation has been found to be reduced/abnormal in recent studies. Tables 8.2 to 8.4 show the results for the modified Nott technique at 20cm obtained with in our sample of Down syndrome subjects. A response with a small lag between 0.50/0.75D was considered a normal result, whereas values smaller than 0.5D were considered a lead of accommodation, and values greater than 0.75D were taken as a considerable lag of accommodation. At the distance of 20cm, values greater than +1D were considered abnormal. The results were analysed as a function of type of refractive error (hyperopes and myopes):

HYPEROPES	TEST DISTANCE	EYE	DISTANCE (cm)	(D)
M.G	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	50	0.5
		UE	50	0.5
J.F	**NOTT** (20cm)	UD	40	2.5
		UE	40	2.5
	NOTT	UD	60	0.83
		UE	60	0.83
I.I	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	R=S
		UE	45	0.25
C.P	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67
	NOTT	UD	60	0.83
		UE	60	0.83
I.S	**NOTT** (20cm)	UD	25	1
		UE	25	1
	NOTT	UD	50	0.5
		UE	60	0.83
C.DF	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	R=S
		UE	40	R=S
C.L	**NOTT** (20cm)	UD	40	2.5
		UE	30	1.67
	NOTT	UD	40	R=S
		UE	40	R=S
G.C	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67

	NOTT	UD	50	0.5
		UE	50	0.5
J.C	**NOTT (20cm)**	UD	25	1
		UE	25	1
	NOTT	UD	70	1.08
		UE	60	0.83
J.J	**NOTT (20cm)**	UD	30	1.67
		UE	30	1.67
	NOTT	UD	50	0.5
		UE	60	0.83
S.S	**NOTT (20cm)**	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	R=S
		UE	40	R=S
D.A	**NOTT (20cm)**	UD	20	R=S
		UE	20	R=S
	NOTT	UD	-	/
		UE	-	/
I.G	**NOTT (20cm)**	UD	30	1.67
		UE	25	1
	NOTT	UD	40	R=S
		UE	40	R=S

Table 8.2: Results for Nott retinoscopy technique at a 20cm and 40cm for hyperopic Down syndrome subjects studied. This table shows the distance were neutral movement was found from the target point, together with the response of the accommodation in dioptres (D). R=S means response at the same point of the target.

SUMMARY FOR HYPEROPIC DS NOTT RESULTS	**NOTT (20cm)**	Within normal limits	0
			0
		Lag	7
			8
		Lead	6
			5
	NOTT	Within normal limits	4
			2
		Lag	4
			5
		Lead	3
			4
		Unstable	1
			1

Table 8.3: Summary for results of Nott modified (20cm) and Nott. It indicates for each test, the number of right and left eyes with normal limit values of accommodation lags or leads of accommodation. Unstable means no conclusion obtained.

MYOPES	TEST DISTANCE	EYE	DISTANCE (cm)	(D)
L.B	**NOTT (20cm)**	UD	40	2.5
		UE	40	2.5
	NOTT	UD	60	0.83
		UE	60	0.83
X.C	**NOTT (20cm)**	UD	20	R=S
		UE	30	1.67
	NOTT	UD	40	R=S
		UE	50	0.5
D.P	**NOTT (20cm)**	UD	30	1.67
		UE	30	1.67
	NOTT	UD	50	0.5
		UE	50	0.5
S.F	**NOTT (20cm)**	UD	20	R=S
		UE	20	R=S
	NOTT	UD	50	0.5
		UE	50	0.5
E.F	**NOTT (20cm)**	UD	25	1
		UE	25	1
	NOTT	UD	45	0.25
		UE	45	0.25
A.N	**NOTT (20cm)**	UD	20	R=S
		UE	20	R=S
	NOTT	UD	50	0.5
		UE	50	0.5
J.L	**NOTT (20cm)**	UD	25	1
		UE	25	1
	NOTT	UD	45	0.25
		UE	45	0.25
M.T	**NOTT (20cm)**	UD	25	1
		UE	25	1
	NOTT	UD	50	0.5
		UE	50	0.5
RX.E	**NOTT (20cm)**	UD	20	R=S
		UE	25	1
	NOTT	UD	40	R=S
		UE	50	0.5

Table 8.4: Results for Nott retinoscopy technique at 20cm and 40cm for myopic Down syndrome subjects studied. This table shows the distance were neutral movement was found from the target point, together with the response of the accommodation in dioptres (D). R=S means response at the same point of the target.

SUMMARY FOR MYOPIC DS NOTT RESULTS	**NOTT (20cm)**	Within normal limits	0
			0
		Lag	5
			7
		Lead	4
			2
	NOTT	Within normal limits	4
			6
		Lag	4
			3
		Lead	1
			0
		Unstable	0
			0

Table 8.5: Summary for results of Nott modified (20cm) and Nott. It indicates for each test, the number of right and left eyes with normal limit values of accommodation lags or leads of accommodation. Unstable means no conclusion obtained.

The results of the lag of accommodation obtained for hyperopes and myopes at the distance of 20cm are similar, as both show a major prevalence for a lag of accommodation at this distance, followed by a smaller proportion of cases with a lead of accommodation and no cases of values within the expected lag for this distance. However, at the distance of 40cm, both hyperopes and myopes show more cases with values within the normal limits and with a similar proportion. Lag of accommodation greater than 0.5/0.75D is also found of in a high proportion of hyperopes than myopes. Also, hyperopes showed more cases of lead of accommodation, while only one myopic eye had a lead of accommodation for this distance, which may suggest overcorrection, although overrefraction was neutral for this subject.

The same analysis was done as a function of sex in order to see if there exist differences in the lag of accommodation between male and female subjects. Table 8.6 show the results for the Nott technique at two different distances (20 and 40cm), obtained for hyperopic and myopic females, and table 8.8 for hyperopic and myopic males:

HYPEROPIC AND MYOPIC FEMALES	TEST DISTANCE	EYE	DISTANCE (cm)	(D)
M.G	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	50	0.5
		UE	50	0.5
L.B	**NOTT** (20cm)	UD	40	2.5
		UE	40	2.5
	NOTT	UD	60	0.83
		UE	60	0.83
I.I	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	R=S
		UE	45	0.25
C.P	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67
	NOTT	UD	60	0.83
		UE	60	0.83
I.S	**NOTT** (20cm)	UD	25	1
		UE	25	1
	NOTT	UD	50	0.5
		UE	60	0.83
S.F	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	0.5
		UE	40	0.5
C.DF	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	R=S
		UE	40	R=S
J.R	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67
	NOTT	UD	50	0.5
		UE	60	0.83
S.S	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	40	R=S
		UE	40	R=S
A.N	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	50	0.5
		UE	50	0.5
M.T	**NOTT** (20cm)	UD	25	1
		UE	25	1
	NOTT	UD	50	0.5
		UE	50	0.5

I.G	**NOTT (20cm)**	UD	30	1.67
		UE	25	1
	NOTT	UD	40	R=S
		UE	40	R=S

Table 8.6: Results for Nott retinoscopy technique at 20 and 40cm for female Down syndrome subjects studied. This table shows the distance were neutral movement was found from the target point, together with the response of the accommodation in dioptres (D). R=S means response at the same point of the target.

SUMMARY FOR DS FEMALES NOTT RESULTS	**NOTT (20cm)**	Within normal limits	0
			0
		Lag	6
			6
		Lead	6
			6
	NOTT	Within normal limits	6
			4
		Lag	3
			5
		Lead	3
			3
		Unstable	0
			0

Table 8.7: Summary for results of Nott modified (20cm) and Nott for females with Down syndrome studied. It indicates for each test, the number of right and left eyes with normal limit values of accommodation lags or leads of accommodation. Unstable means no conclusion obtained.

HYPEROPIC AND MYOPIC MALES	TEST DISTANCE	EYE	DISTANCE (cm)	(D)
J.F	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67
	NOTT	UD	40	R=S
		UE	60	0.83
X.C	**NOTT** (20cm)	UD	20	R=S
		UE	30	1.67
	NOTT	UD	40	R=S
		UE	50	0.5
D.P	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67
	NOTT	UD	50	0.5
		UE	50	0.5
C.L	**NOTT** (20cm)	UD	40	2.5
		UE	30	1.67
	NOTT	UD	40	R=S
		UE	40	R=S
G.G	**NOTT** (20cm)	UD	30	1.67
		UE	30	1.67
	NOTT	UD	50	0.5
		UE	50	0.5
J.C	**NOTT** (20cm)	UD	25	1
		UE	25	1
	NOTT	UD	70	1.08
		UE	60	0.83
E.F	**NOTT** (20cm)	UD	25	1
		UE	25	1
	NOTT	UD	45	0.25
		UE	45	0.25
D.A	**NOTT** (20cm)	UD	20	R=S
		UE	20	R=S
	NOTT	UD	-	/
		UE	-	/
J.L	**NOTT** (20cm)	UD	25	1
		UE	25	1
	NOTT	UD	45	0.25
		UE	45	0.25
RX.E	**NOTT** (20cm)	UD	20	R=S
		UE	25	1
	NOTT	UD	40	R=S
		UE	50	0.5

Table 8.8: Results for Nott retinoscopy technique at 20 and 40cm for males Down syndrome subjects studied. This table shows the distance were neutral movement was found from the target point, together with the response of the accommodation in dioptres (D). R=S means response at the same point of the target.

SUMMARY FOR DS MALES NOTT RESULTS	**NOTT (20cm)**	Within normal limits	0
			0
		Lag	7
			10
		Lead	3
			0
	NOTT	Within normal limits	2
			3
		Lag	5
			4
		Lead	1
			1
		Unstable	1
			1

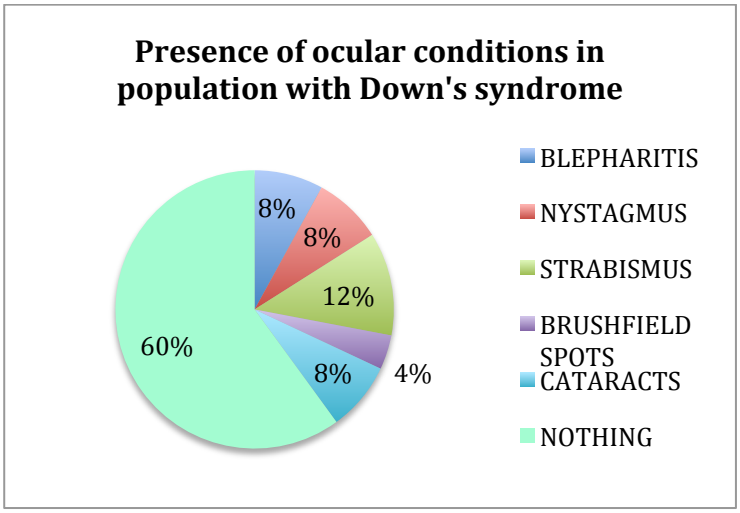
Table 8.9: Summary for results of Nott modified (20cm) and Nott for males with Down syndrome studied. It indicates for each test, the number of right and left eyes with normal limit values of accommodation lags or leads of accommodation. Unstable means no conclusion obtained.

At a distance of 20cm, neither females nor males show a response of accommodation within normal values, but do show a lag of accommodation. Females show the same proportion of cases with a lag and lead of accommodation, while males present more cases with a lag of accommodation than a lead of accommodation. By contrast, at the distance of 40cm, results within the normal limits are more common for both female and male subjects. Also, females show similar proportion of cases with lead and lag of accommodation, while males lag of accommodation is far more common than over accommodation.

Thus, it is suspected that Down syndrome subjects in our small sample present an inability to accommodate accurately at the distance of 20cm, regardless of sex or the type of refractive error, but can accommodative more accurately at the distance of 40cm. Moreover, male myopes show less proportion cases to over accommodate than hyperopic female subjects.

8.4 OCULAR HEALTH

The presence of ocular conditions was also analysed in our sample of Down syndrome subjects. Conditions like blepharitis, nystagmus, strabismus, brushfield sports and cataracts were recorded. Graph 8.5 shows the results obtained for type and frequency of ocular conditions present in our sample of Down syndrome subjects.



Graphic 8.5: Type and frequency of ocular conditions present in our sample of Down syndrome subjects.

A 60% of the sample studied did not have any ocular condition. Among those who did (40%), strabismus was the most common ocular condition found, usually associated with nystagmus. In most cases the strabismus found disappeared with refractive correction, which suggests an accommodative strabismus type. Two of the subjects with strabismus had significant refractive error (+2.5D /+3.5D and +5D/+5D). Another subject with strabismus only had little myopia (-0.25D) in both eyes, which suggests that the strabismus was not related to refractive error.

Blepharitis, as well as cataracts, was also present in some cases. Due to the limited instrument resources available for assessment, which characterises this type of home/centre visits screening examination, the existence of cataracts was considered when there was media opacies seen with retinoscopy and associated to poor visual acuity. Brushfield spots were seen in only one subject who was 6 years old.

8.5 COLOUR VISION AND STEREOPSIS

8.5.1 Colour vision

Colour vision deficiencies were assessed using the Ishihara 38 plates test. Normal results were considered when all numbers plates presented were seen correctly, abnormal results when with any mistake in recognizing a number and correct result. The results obtained are exposed in the next table (8.9):

COLOUR VISION	SEX	RESULTS
M.G	**F**	Normal
J.F	**M**	Normal
L.B	**F**	Normal
X.C	**M**	Normal
D.P	**M**	Normal
I.I	**F**	Normal
C.P	**F**	Normal
I.S	**F**	Normal
S.F	**F**	Correct except 33,34,36,37
C.DF	**F**	Normal
C.L	**M**	Correct except 33
G.G	**M**	Correct except 32
J.C	**M**	Normal
J.R	**F**	Normal
E.F	**M**	Normal
S.S	**F**	Normal
D.A	**M**	Normal
A.N	**F**	Normal
J.L	**M**	Normal
M.T	**F**	Normal
RX.E	**M**	Normal
I.G	**F**	Correct except 33

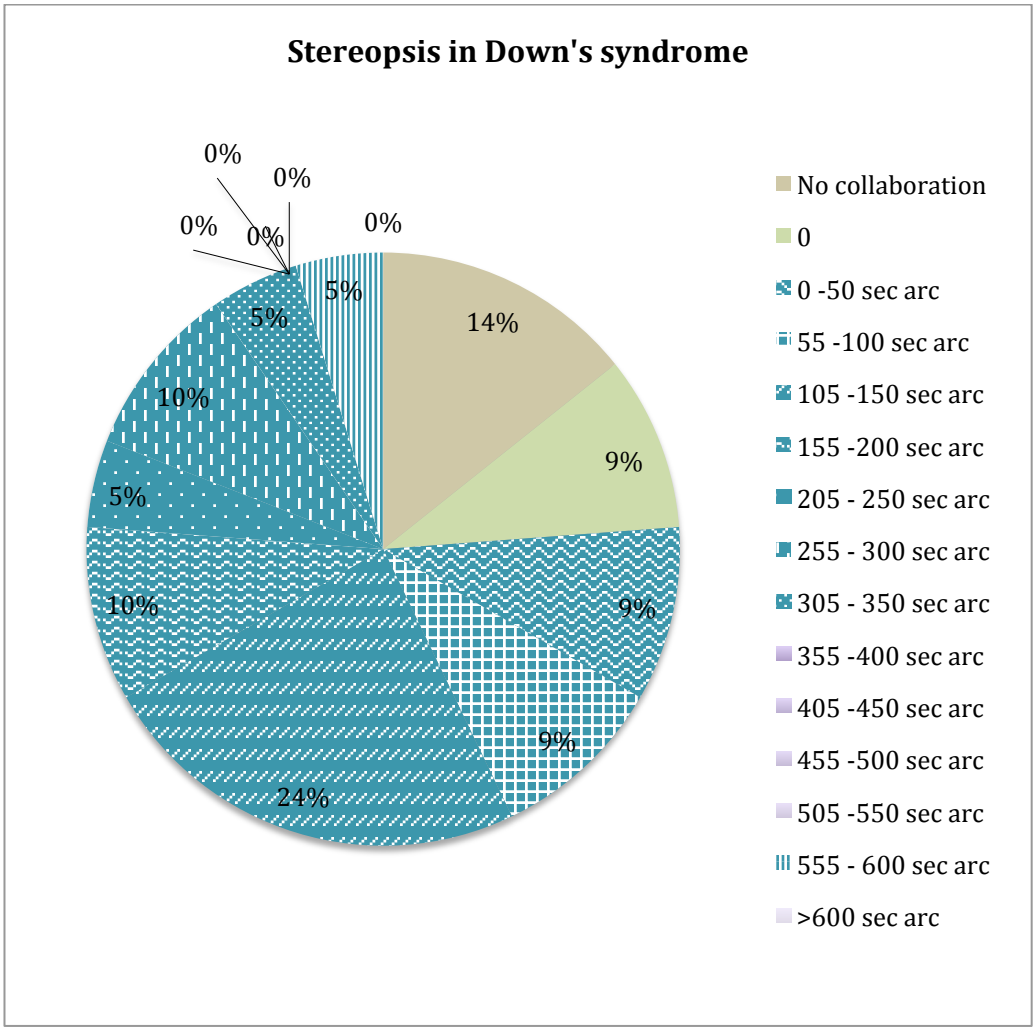
Table 8.9: Results for the Ishihara test colour vision deficiency for each subject examined. Normal results were assigned when all numbers were seen correctly, abnormal results when numbers were not seen. Finally, plates failed but the rest of the numbers where seen correctly, it was noted too.

As it can be seen in table 8.9, all subjects presented normal colour vision. However, 4 out of 22 subjects had difficulties with some of the plates shown. The most common card failed was when asking for card number 32 and 33.

A failure in plates 32 and 33 mean colour blindness if they don't trace the line correctly or they don't see any line. A failure in plates 34 would mean a deficiency of red-green vision if they connect the bluish-green and purple, or a total colour blindness or weakness if they do not see any line, while a failure in plates number 36 and 37 with a connection of the line of purple and bluish-green would mean a failure in red-green colours vision, and blindness if they don't see any line.

8.5.2 Stereopsis

Stereoacuity was assessed using the Frisby stereotest. The average of stereoacuity is represented in the following graphic (8.6), which shows the proportion of Down syndrome subjects and value of stereoacuity:



Graphic 8.6: Existance and degree of stereopsis determined with Frisby stereotest in a sample of corrected Down syndrome subjects. Stereoacuity is measured in seconds of arc.

Stereopsis was present in more than ¾ of the sample (77%). Those subjects who did not cooperate were the youngest of the sample (6, 14 and 15 years of age). A 9% of subjects did not demonstrate stereopic vision. These subjects did not have high refractive error nor anisometropia. The rest of the sample (light green) showed stereoacuity. As it can be seen in the graphic 8.6, a large proportion of the sample (24%) presents stereoacuity between 105-150 sec arc, followed by 10% stereacuities between 155-200 sec arc and 255-300 sec arc (10%). No subjects responded to stereoacuities between 355 to 550 sec arc and higher than 600 sec arc, and only one subject had stereoacuity of 600 sec arc and another of 340 sec arc.

9 DISCUSSION AND CONCLUSION

This discussion aims to compare the results obtained in the literature and the results obtained in our small study, in order to come to conclusions about different aspects of ocular health and visual function characteristics of Down syndrome population.

It has been shown that visual acuity has a similar development with normal subjects until the age of infancy, but then falls below the normal limits (J.A. Little et al.; 2007). Subjects with DS have shown poorer visual acuity in comparison to their matched aged normal subjects (JA Little et al.; 2007, Courage et al.; 1994, Haugen et al; 2001, Woodhouse et al 1996). Courage et al.; (1994) also found no correlation between visual acuity and refractive error similarly did our study, which showed that some subjects could have good or bad visual acuity regardless of the spherical equivalent error (SER).

J.A. Little et al.; (2007) found that the mean visual acuity result is 6/13 (0.33 ± 0.18 in decimal scale). The mean results in the present study was 0.29(± 0.10) visual acuity in decimal scale for the right eye, 0.29(± 0.12) for the left eye and a mean of 0.50(± 0.17) in decimal scale for binocular vision. No difference seems to be apparent between females and males. Furthermore, visual acuity doesn't seem to reach normal limits even with full correction. The occlusion for the visual acuity test was done better with hands rather than with the occluder. When none of the symbols from the first raw were seen, a possible reason for it was a matter of insecurity and after giving a matching card the results improved considerably reaching values of decimal visual acuity as good as 0.63 decimal.
No subject had a monocular visual acuity better than 0.6 for the left eye, but there was a subject with visual acuity smaller than 0.1 (0.06). This subject also presented poor visual acuity for the right eye. No improvement was found binocularly with this subject. A cataract seen in the right eye would explain the bad visual acuity for this eye, but not for the left eye. Also this subject was not wearing any glasses and that is why he was corrected with trial frame with +0.5D in the right eye. Maybe that could influence the result, as it was not well corrected at all. No strabismus was found. More investigation should have had to be done in this subject.
The reasons for the poor visual acuity are still discussed. It is agreed by Little et al.; (2007), Ahmad et al.; (1976), Liza-Sharmini AT et al.; 2006) that poor optical quality has implications for retinal and cortical image quality; others such as F.M. John et al.; (2004), Weijerman (2010) and Ellingston (1986) suggest that there are differences in the visual cortices of the brain in Down's syndrome which lead to the poor visual acuity (5.1.1.)

The present study did not asses the Contrast Sensitivity Function of the group of Down syndrome subjects studied, due to time constrains, however this visual function has been reported to be altered, as well as in this population. J.A. Little et al.; (2007) and Courage et al.; 1997 agreed that CSF for Down's syndrome subjects follows a similar pattern as a CSF for normal subjects, but the curve follows a pattern under normal

limits and it happens at all ages. Bigger differences are seen at higher spatial frequencies (5.1.1). Suttle and Tuner.; (2004) suggested a neural basis as a reason for it.

One of the most discussed aspects of vision in Down syndrome population is the high incidence for refractive errors. Several authors (M. AlBagdady et al.; 2010, T. Watt et al.; 2014) have demonstrated a considerable difference between the prevalence of refractive error in population with Down syndrome compared to normal subjects, which it tends to be greater with aging. This seems to be obvious by the preschool age, with 50% of incidence in Down's syndrome compared an incidence of 5.8% in normal children (table 5.1). Almost all studies (T. Watt et al.; 2014, Courage et al.; 1997, Woodhouse et al.; 1993, Al-Bagdady; 2011, Ljubic A et al; 2011) agree in the failure of emmetropization in Down syndrome subjects although there is no firm explanation for it. Due to the failure of emmetropization, the common refraction in new-borns and infancy remains fairly stable instead of reducing as it happens with normal subjects. This could not been proved in our project as it is needed to follow a continued study and the sample is small to make comparisons with matching controls.
The incidence of myopia and hyperopia in Down syndrome is greater, and hyperopia is more common than myopia (Woodhouse et al; 1997). This is in line with our findings in the clinical assessment in Fundació Down de Lleida, where I obtained a higher proportion of hyperopia in Down syndrome subjects, greater than myopia, although it was seen that for ages between 19 to 29 myopia was predominant. This might have been due to the small sample. I also observed that female subjects with Down syndrome tend to be more hyperopic than male. In our study cases of high hyperopia and high myopia were rare.

It is suggested that the spread of refractive errors is larger than normal population. J.M Woodhouse et al.; (1993) demonstrated that the range for refractive errors in Down syndrome goes from -12D to +3.5D. In our small sample of Down syndrome subjects, the spread of refractive error for hyperopia is about $+2.39 \pm 1.65$D and $+2.879 \pm 1.52$D for the right and left eye, while the mean myopic refractive error (SER) is -3 ± 1.85D and -2.709 ± 1.69D respectively. Only few subjects presented high refractive error (SER) greater than +5D or -6D. All cases were all females.
Several studies suggest that myopia becomes more common in Down syndrome up to adolescence (J. Puig et al.; 2002). The present study only had two adolescents and they were not myopes.

Failure of emmetropization also affects astigmatism, which does not show the typical reduction pattern found in normal subjects. Primary school Down syndrome children show increased levels of astigmatism, while normal primary school children normally don't (J.M. Woodhouse; 1999). The most frequent type of astigmatism is with-the-rule astigmatism and oblique astigmatism (Al-Bagdady; 2010), which agrees with the results of our study. In the present study, astigmatisms ≥ 0.75DC were considered relevant and values of astigmatism considered irrelevant (<0.75DC). The most predominant type were with-the-rule and oblique astigmatisms (2/3 of the sample), in agreement with the literature. However, 1/3 of the sample showed no astigmatism or irrelevant. The mean astigmatism value found in our small sample was 2DC.

The abnormal palpebral fissure of the eyelid is the major aetiological factor for corneal astigmatism (Watt; 2014, Haugen; 2001, M. Al-Bagdady; 2011) and the angle of the corneal astigmatism has been correlated with the palpebral fissure (Read; 2007, Shafiro; 1985). In the present study, it has been also seen correlation with the slanted palpebral fissure for some of the cases with oblique astigmatism found. In the right eye, 4/14 eyes with astigmatism followed the pattern of the axis at 135º±10º caused by the shape of the eyelid, and 2/13 left eyes presented oblique astigmatism (45º±10º). However, no conclusive results can be drawn from these data, due to the small sample size.
Furthermore, astigmatism was found in almost all subjects of our sample. Several studies suggest that astigmatism increases with age, however this is something that could not be studied in our samples since it would require an on going longitudinal study.

Accommodation is still the most discussed and studied visual function in Down syndrome as there is no reasonable explanation for the under accommodation of these subjects and also for the large affection that has over Down's syndrome.
This was seen during dynamic retinoscopy at two distances of 20cm and 40cm, in which 48% of the samples showed to under accommodate and results within the normal limits were more frequent at 40cm distance. Over 70% of Down syndrome subjects under accommodate at near distances (J.M. Woodhouse et al.; 2000). Cregg et al.; (2001), Olav H. Haugen et al.; (2001), which is in accordance with our results demonstrated large under accommodation at all distances tested, which increases with the accommodative demand and decreases with age. The present study found no differences in the prevalence of lag of accommodation between myopes and hyperopes.
The limit of accommodation for Down's syndrome lays within 6-8D at 16-12.5cm while normal young subjects show accurate accommodation of 6-4D at 16-25cm (Cregg et al.; 2001, T. Watt et al.; 2014, Rouse et al.; 1984).

A value greater than 1D for lag of accommodation is considered an abnormal value for accommodation response. However, in Down's syndrome 55% of subjects have lag of accommodation greater than 1D at working distances of 20-30cm (Olav H. Haugen et al.; 2001). These results agree with the results found in the present study, where 57.69% of the sample of Down syndrome subjects examined showed lag of accommodation of 1.31D. Accommodative response does not improve with full correction.
The number of subjects with lag of accommodation between 0.5-0.75DC is greater in the hyperopic group compared to the myopic group of subjects. As a function of sex, males showed more lag of accommodation than females, and female subjects showed more results within the normal limits for the distance of 40cm than males.
It can be concluded that Down syndrome subjects present inability to accommodate accurately at closer targets, regardless of sex and type of refractive error. This was also seen in the subjects observed during my collaboration with J.M. Woodhouse and her team at the exchange at Cardiff University of Optometry.

Reasons for poor amplitude of accommodation and under accommodation is still not known, but it has been shown in Haugen O.H.; (2001), HØvding and Eide.; (2001) that on average, the lens is thinner in the centre and the power of the lens is lower (12.70±2.36D) compared to 19.480±1.24D in normal subjects. The parasympathetic nervous system has also been suggested as a reason for inaccurate ability to accommodate (Haugen and HØvding.; 2001)but all conclude that need further investigation.

The assessment carried out at the Fundació Down Lleida also checked for ocular anomalies. It has been reported that Down syndrome subjects suffer from conditions like cataracts, brushfield spots, blepharitis, strabismus and more.
The sample of Down syndrome subjects studied at Fundació Down Lleida showed a 60% of cases that did not present ocular anomalies and a 40% of cases that did. Among the ocular anomalies or conditions found strabismus was the most prevalent (12%) followed by blepharitis, nystagmus and cataracts (8%), and the least, brushfield spots (4%).

Most of the cases observed with strabismus were accommodative strabismus and most disappeared with correction, although there are studies, which demonstrated that accommodation is not the only factor that causes strabismus, as myopia could not explain the presence of strabismus.
Those cases with strabismus were low myopic (-0.25 and -1.5D) and one with high hyperopia (+5D). This shows that accommodation is not the only factor that may cause strabismus. Some studies have demonstrated that strabismus is found in more prevalence in preschool Down syndrome subjects than in Down syndrome infants and that the possibility to suffer from strabismus increases with age. In normal subjects the chance of a child presenting strabismus increases in the first two years of age and after decreases significantly.
We found no cases of strabismus in the primary school Down syndrome children studied.

Only few studies have done research about stereopsis and colour vision in Down's syndrome. This study, few subjects presented good stereoacuity (5%), but most of them (24%) reached a threshold of 100-150 sec arc. Only 9% of the subjects had good stereopsis (0-50 sec arc).
In this study, literature has shown that colour deficiencies are more common in Down syndrome subjects. Up to a 23% of cases (Adams et al; 1993, Lowe 1949, Mills; 1984) with a higher prevalence of protanopia, has been reported in Down syndrome population compared to normal subjects. Although three subjects failed plate 33 and one subject plate 32 of the Ishihara 38 plates test, which could be attributed to colour vision deficiencies the rest of the plates were correctly identified, which may indicate lack of attention from the subjects. Card number 34, 36 and 37 also appeared to be difficult for one subject, but probably was due to the fatigue.

From the results obtained for the subjects studied at the Fundació Down Lleida, I conclude that subjects have their visual needs were better looked after than I expected. All subjects were correctly or nearly well corrected with spectacles and they

were living a nearly independent life, which helped with the understanding of the tasks demands carried out and were alert and active. Lot of collaboration was received from their part and this is reflexed in the results.

The goals and requirement for learning the techniques and methods needed to deal with this population were achieved with my participation in several screening and full assessment of Down syndrome children and mentally disabled subjects, first in Cardiff University School of Optometry and later on with the Down syndrome sample assessed at Fundació Down Lleida. I could also learn how to interact with Down syndrome and mentally disabled population, which improved my professional skills and my English language.

Although my findings are only anectodical given the small sample studied they have allowed my to compare with other studied and demonstrate what I found in my sample studied was in line with other authors.

To conclude, the thorough literature review carried out and my personal experience through the sample of Down syndrome subjects assessed and the short residency at Cardiff University School of Optometry, has given me the experience and practise necessary to manage this population.

10 BIBLIOGRAPHY

- A.J. Adams, C.R. Cavonius, S.D. Dain, G. Haegerstrom-Portnoy, K. Kitahara, K. Knoblauch, A. Kurtenbach, B.B Lee, J. Mollon, J. D. Moreland, J. Pokorny, L.T. Sharpe, H.A. Sperling, W.H. Swanson, E. Zrenner. Edited by B. Drum. "Colour Vision Deficiences XII". Docum Opthal Proceedings Series 57. July 1993.
  6. J. Pérez-Carpinell, M.D. de Fez and V. Climent.
  Visual defects in subjects with Down's syndrome. Pp. 43-47.

- Al-Bagdady M, Murphy PJ, Woodhouse JM. "Development and distribution of refractive error in children with Down's syndrome". Br J Ophthalmol 2011; 95; pp. 1091–1097.

- Al-Bagdady M., Paul J. Murphy, Woodhouse JM. "Develoment and distribution of refractive error in children with Down's syndrome." School of Optometry and Vision Sciences, Cardiff Univerity. 7th October 2010.

- Berk A.T, Saatci AO, Ercal MD. "Ocular findings in 55 patients with Down's syndrome". Ophthalmic Genet 1996; 17; pp. 15-19.

- B. Haargaad, H.C. Fledelius. "Down's syndrome and early cataract". Br J. Ophthalmol 2006; 90, pp. 1024-1027.

- Bittles A.H, Glasson E.J. "Clinical, social and ethical implications of changing life expectancy in Down syndrome". Dev Med Child Neurol 2004; Vol 46; pp. 282–286.

- Bhatti T.R, Dott M, Yoon PW et al. "Descriptive epidemiology of infantile cataracts in metropolitan Atlanta, GA, 1968-1998". Arch Pediatr Adolesc Med 2003; 157; pp. 341-7.

- Courage ML, Adams RJ, Hall FJ. "Contrast sensitivity in infants and children with Down syndrome". Vision Res 1997; Vol 37; pp. 1545-1555.

- Creavin A, Brown R. "Ophtlamic Abnormalities in children with Down syndrome". J Pediatr Ophtalmol Strabismus 2009; 46; pp. 76-82.

- Da Cunha RP, Moreira JB. Ocular findings in Down's syndrome. Am J Ophthalmol 1996; 122; pp. 236–44.

- Da Cuna R.P, Moreira JBDC. "Ocular findings in Down's syndrome". Am J Ophtalmol 1996; 122; PP. 236-244.

- Deng L, Gwiazda JE. “Anisometropia in children from infancy to 15 years”. Invest Ophthalmol Vis Sci 2012; Vol 53; pp. 3782–3787.

- Don C. Van Dyke, David J. Lang, F. Heide, S. Van Duyne, M.J. Soucek. “Clinical Perspectives in the management of Down syndrome”. Sprinfer-Verlag New York Inc, 1990.

- Down’s syndrome association medical series. “Nº eye problems in children with Down’s syndrome”.

- Esparza, H.; Garcia, M. P. “Desarrollo de la eficiencia visual”. Universidad Autónoma de Aguasacalientes, 2014.

- “Eye problems in children with Down’s syndrome: notes for parents and carers”. Down’s Syndrome Association Medical Series, August 2007.

- Fabrizio Bonci, Dip. Optom (ITA), MCOtopm, Luigi Lupelli, Dip. Optom (ITA), FAILAC, FIACLE, FBCLA. “Retinoscopy in infancy: cycloplegic versus non-cycloplegic”. Ot CET continuing education and training. Academy for eyecare excellence, Ciba Vision. C-18551 O/D. 6/4/2012

- Ffion M. John, Nathan R, Bromham, J. Margaret Woodhouse, and T. Rowan Candy.” Spatial Vision Deficits in Infants and Children with Down syndrome”. Inv Ophthalmol & Vis Sci; 2004; 45, Nº 5; pp. 1566- 1571.

- Fierson W.M, Van Dynke DC, Lang DJ, Heide F, et al, eds. “Clinical perspectives in the management of Down’s syndrome”. Ophthalmological aspects. Springer-Verlag; 1990; pp. 26–54.

- Gardiner PA. “Visual defects in cases of Down’s syndrome and other mentally handicapped children”. Br J Ophthalmol 1967; 57; pp. 469-74.

- “Guía oftalmológica del síndrome de Down”. Down España. Fundació Catalana Síndrome de Down. Dr. Javier Puig Galy, Dra. Alicia Galán Terraza. Departamento de Oftlamología, Centro Médico Down de la Fundació Catalana Síndrome de Down. Dr. José Mª Borrel Martínez, asesor médico de Down España.

- Haugen OH, Høvding G, Lundstrom L. “Refractive development in children with Down’s syndrome: a population based, longitudinally study”. Br J Ophthalmol 2001; Vol 85; pp. 714-719.

- Haugen OH, Høvding G, Eide GE. “Biometric measurements of the eyes in teenagers and young adults with Down syndrome”. Acta Ophthalmol Scand 2001; 79; pp. 616–625.

- Haugen OH, Høvding G; “Strabismus and bincoular function in children with Down syndrome. a population-based, longitudinal study”. Acta Ophtalmol Scand 2001; 79; pp. 133-139.

- Igersheimer J. “The relationship of lenticular changes to mongolism”. Trans Am Soc Ophthalmol Otolaryngol Allergy 1951;49; pp. 595–624.

- Javier Puig Galy, Alicia Galán Terraza, Manuel Romera Becerro. “Caso Clínico-Tortículis ocular en el niño con síndrome de Down”. SD, Revista Médica Internacional sobre el Síndrome de Down. 2006: vol 10, núm1, pp. 8-12

- Julie-Anne Little, J. Margaret Woodhouse, Jan S. Lauritzen and Kathryn J. Saunders. “The impact of Optical Factors on Resolution Acuity in Children with Down syndrome.” Inv Ophtoalmol and vis Sci, September 2007; 48, 3995-4000.

- J. M. Woodhouse, Jennifer S. Meades, Susan J. Leat and Kathryn J. Saunders. “Reduced Accommodation in Children with Down syndrome. Investigate Ophthalmology & Visual Science, June 1993, Vol 34, Nº 7 pp. 2382-2387.

- J. M. Woodhouse, PhD, Valerie H. Pakeman, BSc, Mary Cregg, DipOptom, Kathryn J. Saunders, PhD, Margaret Parker, Med, William I. Fraser, MD, Prema Sastry, MB, and Sandra Lobo, MB. “Refractive errors in young children with Down Syndrome”. Optometry and Vision Science, October 1997. Vol. 74, Nº 10.

- J. M. Woodhouse, May Cregg, Helen L. Gunter, Daniel P. Saunders, Valerie H Pakeman, Margaret Parker, William I. Fraser, and P. Sastry. “The effect of Age, Size of Target, and Cognitive Factors on Accommodative Responses of Children with Down syndrome”. Inv Ophthalmol & Vis Sci; 2000; 41 Nº9, pp. 2479-2485

- J. M. Woodhouse. “Vision in children with Down syndrome: a research update”. Down’s syndrome Vis Research Unit, School of Optom & Vis Sci, Cardiff University. 2005, Pp. 87-89.

- J. M. Woodhouse J.M, Pakeman VH, Cregg M, Saunders KJ, Parker M, Fraser WI, Sastry P et al. “Refractive errors in young children with Down syndrome”. Optom Vis Sci 1997; Vol 74; pp. 844–851.

- J. M. Woodhouse JM, Pakeman VH, Saunders KJ, Parker M, Fraser WI, Lobo S, Sastry P. “Visual acuity and accommodation in infants and young children with Down’s syndrome”. J Intelled Disabil Res, 1996; 40; pp. 49-55.

- Joanquin Pérez-Carpinell, Pacual Capilla, Carlos Illueca, Jesus Morales. “Vision defects in albinism. Optometry and vision science”. American academy of optometry, 1992. Vol 69, Nº 8, pp. 623-628.

- John FM, Bromham NR, Woodhouse M, Candy TR. “Spatial vision deficits in infants and children with Down syndrome”. Invest Ophthalmol Vis Sci 2004;Vol 45; pp. 1566–1572.

- J. Puig, E. Estrella, A. Galán. “Ametropía y estrabismo en el niño con syndrome de Down”. SD. Revista médica Internacional sobre el syndrome de Down. 2002: vol 6, núm 3, pp. 34-39.

- Karlica D, Skelin S, Culic V, Galetovic D, Znaor L, Karlica H, Pavelic J. “The ophthalmic anomalies in children with Down syndrome in Split-Dalmatian County”. Coll Antropol 2011; Vol 35; pp. 1115–1118.

- Kathryn J. Saunders, PhD, MCOptom; J. Margaret Woodhouse, PhD; and Carol A. Westall, PhD, FCOptom.” The modified Frisby Stereotest”. J Pediatr Ophthalmol Strabismus 1996; 33; pp. 323-327.

- Kathryn J. Saunders, PhD, MCOptom; J. Margaret Woodhouse, PhD; and Carol A. Westall, PhD, FCOptom. “The modified Frisby Stereotest”. J Pediatr Ophthalmol Strabismus 1996; Vol. 33; pp. 323-327.

- Kleinstein RN, Jones LA, Hullett S, Kwon S, Lee RJ, Friedman NE, Manny RE et al. “Refractive error and ethnicity in children”. Arch Ophthalmol 2003; Vol 121; pp. 1141–1147.

- Lea Averbuch-Heller, M.D., Louis F. Dell’Osso, PhD, Jonathan B. Jacobs, M.S., and Bernd F. Remler, M.D. “Latent and Congenital Nystagmus in Down Syndrome”. Joun of Neuro-Ophthalmol; 1999, Vol 19(3); pp. 166-172.

- Lejeune, L., Gautier, M. & Turpin. “Les chromosomes humainsen culture de tisshu.” Comptes Rendus de l'Academie des Sciences, 1959; 248, pp. 602-603.

- Ljubic A, Trajkovski V, Stankovic B. “Strabismus, refractive errors and nystagmus in children and young adults with Down syndrome”. Ophthalmic Genet 2011;Vol 32; pp. 204-211.

- Mª Auxiliadora Robles Bello. “Incidencia y prevalencia del syndrome de Down”. Revista syndrome de Down. Junio 2007, Vol 24.

- Mary Cregg, J. Margaret Woordhouse, Valerie H. Pakeman, Kathryn J. Saunders, Helen L. Gunter, Margaret Parker, William I. Fraser and Prema Sastry. “Accommodation and Refractive error in children with Down syndrome: cross-section and Longitudinal Studies”. IOVS, January 2001, Vol 42, Nº 1

- Mary L. Courage, Russell J. Adams, Elizabeth J. Hall. “Contrast sensitivity in infants and children with Down syndrome”. Vision Res 1997, Vol. 37, Nº 11, pp. 1545-1555.

- Merin S, Crawford J.S. “The etiology of congenital cataracts. A survey of 386 cases”. Can J Ophthalmol 1971;Vol. 6; pp. 178–82.

- Michel E. Weijerman and J. Peter de Winter. “Clinical practice. The care of children with Down syndrome”. Eur J Pediatr. December 2010; 169(12); pp. 1445–1452.

- Millis EA. “Ocular findings in children”. Lane D, Stratford B, eds. “Current Approaches to Down’s syndrome”. London: Holt, Rhinehart and Winston, 1985: 103-19.

- M. Madison Slusher, M.D., Peter R. Laibson, M.D. and Robert D. Mulberger, M.D. “Acute Keratoconus in Down’s syndrome.” American Journal of Ophthalmology, Volume 66, 6, December 1968, 1137–1143.

- Mohammad Al-Bagdady, Paul J. Murphy, J. Margaret Woodhouse. “Develoment and distribution of refractive error in children with Down’s syndrome”. School of Optometry and Vision Sciences, Cardiff Univerity. 7th October 2010

- Mohammad Al-Bagdady, Ruth E. Steward, Patrick Watts, Paul J. Murphy and J. Margaret Woodhouse. “Bifocals and Down’s syndrome: correction or treatment?”. Ophthal. Physiol. Opt. 2009. Vol 29, pp. 1-6.

- M. Madison Slusher, M.D., Peter R. Laibson, M.D. and Robert D. Mulberger, M.D. “Acute Keratoconus in Down’s syndrome”. American Journal of Ophthalmology, December 1968, Vol 66 (6), pp. 1137–1143.

- N.R. Bromham, J.M. Woodhouse, M. Cregg, E. Webb, W.I. Fraser. “Heart defects and ocular anomalies in children with Down’s syndrome”. Br Ophthalmol. December 2002; 86(12); pp. 1367-1368.

- Olav H Haugen, Gunnar HØvding, Isa Lundström. “Refractive development in children with Down’s syndrome: a population based, longitudinal study”. Department of Ophthalmology, Haukeland University Hospital, Bergen, Norway. British Journal of Ophthalmology, June 2001, Vol. 85, Nº6, pp. 714-719.

- Olav H. Haugen and Gunnar HØvding. “Strabismus and binocular function in children with Down syndrome. A population-based, longitudinal study”. Department of Ophthalmology, Haukeland University Hospital, Bergen, Norway. Acta Ophthalmol. Scand. 2001. Vol 79, pp. 133-139.

- Pearce FH, Rankine R, Ormond A. W. “Notes on twenty-eight cases of Mongolian imbeciles: with special reference to their ocular condition”. BMJ 1910;2: 186–90.

- Pérez-Carpinell J, de Fez MD, Climent V. ”Vision evaluation in people with Down’s syndrome”. Ophthalmic Physiol Opt. 1994 Apr;14(2):115-21.

- Pires da Cunha R, Belmiro de Castro Moriera J. “Ocular findings in Down’s syndrome”. Am J Ophthalmol 1996; 122: pp. 236–44.

- “Programa español de salud para personas con syndrome de Down”. FEISD (Federación española del syndrome de Down). Ministerio de Sanidad y comunidad.

- “Programa de Salut per a persones amb Síndrome de Down”. Down Catalunya. Coordinadora Síndrome de Down de Catalunya (CSDC). 2005; pp. 24.

- Rahi J.S, Dezateux C. “Congenital and infantile cataract in the United UK: underlying or associated factors”. British Congenital Cataract interest Group Invest Ophthalmol Vis Sci 2000; 41; pp. 2108-14.

- Raimundo Jiménez Rodríguez, Rosario González Anera, José Ramón Jiménez Cuesta. “Actualización optométrica pediátrica: función acomodativa (II)”. Gaceta óptica, Diciembre 2013-14.

- Randall T. Jose. “Vision subnormal”. Centro de rehabilitación visual de la O.N.C.E. Organización Nacional de ciegos españoles. 1988; pp. 30.

- Roizen NJ. Mets MB, Blondis TA. “Ophthalmic disorders in children with Down syndrome”. Dev Med Child Neurol 1994; 36; pp. 594-600.

- Rosenfield,Nicola Logan,Keith H. Edwards. “Optometry: Science, Tecnhiques and Clinical Management”. Clinical asssessment of accommodation. Chapter 15.. Elsevier, 2009; pp. 231.

- Rouse MW, Hutter RF, Sluftlett R. “A normative study of the accommodative lag in elementary school children”. Am J Optom Physiol Opt 1984; 61, 693-697.

- Ruth E. Stewart, PhD, BSc, MOptom, J. Margaret Woodhouse, PhD, BSc, FSMC, Mary Cregg, PhD, DipOptom, and Valerie H. Pakeman, BSc, MOptom. “Association between Accommodative accuracy, Hypermetropia, and Strabismus in Children with Down’s Syndrome”. School of Otopmetry and Vision Sciences, Cardiff Univerity, Cardiff, United Kingdom. February 2007. Vol 84, Nº 2, pp. 149-155.

- Shapiro MB, France TD. “The ocular features of Down’s syndrome”. Am J Ophthalmol 1985; 99; pp. 659– 663.

- Suttle CM, Turner AM. “Transient pattern visual evoked potentials in children with Down’s syndrome”. Ophthalmic Physiol Opt 2004; 24: pp. 91–99.

- Tanisha Watt, Kenneth Robertson, Robert John Jacobs. “Refractive error, binocular vision and accommodation of children with Down syndrome”. Clinical and Experimental Optometry, 21 September 2014.

- Wagner RS, Caputo AR, Reynolds RD. Nystagmus in Down’s syndrome. Ophtalmology 1990,97; 1439-44.

- Wesson MD, Maino DM. “Occulovisual findings in children with Down’s syndrome, Cerebral Palsy and mental retardation without specific etiology”. In: Maino DM, ed. Diagnosis and Management of Special Popilations. New York: Mosby, 1995: pp. 17-54.

- Wesson M.D, Mann K.R, Bray N.W (1990). “A Comparison of cycloplegic refraction to the near retinoscopy technique for refractive error determination”. J Am Optom Assoc. 1990; 61; pp. 627-38.

- Wirth M.G, Russell-Eggitt I.M, Craig J.E et al. “Aetiology of congenital and paediatric cataract in an Australian population”. Br J Ophthalmol 2002; Vol. 86; pp. 782–6.

10.1 WEBGRAPHY:

- “Supporting people with Down’s syndrome throughout their lives”. Down’s syndrome association. Nº 1061474.

- “ What is Down syndrome?”. National Down syndrome society. 2012.

- “Down syndrome”. MedlinePlus. Trusted information for you. U.S. Department of Health and human services. 9th May 2014.
www.nlm.nih.gov

- “What is Down syndrome?”. Down syndrome International. Improving Life for people with Down syndrome; 2014. Nº 1091843.
www.ds-int.org

- “Cataracts”. VisionWeb; 2010.
www.visionweb.com

- “HOTV”. Lions of Virginia 24D.
www.lions-of-virginia-24d.org

- “Frisby stereotest”. Frisby stereotest UK.
  www.frisbystereotest.co.uk

- “Chromosomal abnormalities”. Virtual genetics education centre. University of Leicester.
  www2.le.ac.uk

- “Chromosome abnormalities”. National human genome research institution. Advancing human Health through genomics research. June 2015.
  www.genome.gov

- “Down syndrome”. Learn Genetics. Genetisc science learning centre. University of Utah Health Sciences; 2015.
  www.learn.genetics.utah.edu

- Google images

- National Down syndrome society (NDSS), “What is Down syndrome?”; 2012
  www.ndss.org

- Down’s syndrome association Medical Series. “Eye problems in children with Down’s syndrome. Ntes for parents & carers”; 2007.
  www.downs-syndrome.org.uk

11 ANNEXS

Annex I: Information sheet for the patient and informed consent form.

- CONSENT FORM

Títol de l'estudi: "**Aspectes visuals de la població amb Síndrome de Down"**

Sr/Sra .....com a pare/mare / tutor legal de....., amb DNI..... domicili ..... i ..... anys d'edat, dóna el seu consentiment per que en/na ..... participi en aquest estudi.
Estic d'acord en que les dades relatives a aquest estudi siguin guardades, processades electrònicament i trameses, pel qual dono el meu consentiment per a que es reveli la informació necessària recollida durant l'estudi per a que pugui ser processada i difosa a la comunitat científica, sense que en cap moment sigui revelada la identitat, ja que els drets de confidencialitat queden protegits.
He rebut suficient informació sobre l'estudi i tots els meus dubtes i preguntes han sigut aclarits.

_____, _____ de_____ de _____

Firma del pare/mare/ tutor legal

Confirmo que he explicat al pacient el caràcter i el propòsit del projecte de la investigació.
Firma del investigador

- INFORMATION SHEET FOR THE PATIENT

Títol de l'estudi: "**Aspectes visuals de la població amb Síndrome de Down"**

Aquest estudi correspon a un treball de final de grau (TFG) dels estudis de Grau en Òptica i Optometria.
L' interès d'aquest treball és estudiar les característiques visuals més rellevants de les persones amb la síndrome de Down. Per això, es determinarà l'error refractiu que predomina i la resposta acomodativa en una mostra de població amb la síndrome de Down.

Les proves que es realitzaran són:

- Determinació de l'error refractiu amb auto-refractòmetre. És un aparell optomètric que permet calcular l'error refractiu de forma automàtica, per mitjà de l'observació d'un objecte a la distància.
- Retard acomodatiu. És una prova que ens permet determinar quina acomodació exerceix el pacient en una determinada distància i poder observar si presenta hiperacomodació o hipoacomodació. Es realitzarà a tres distàncies diferents.
- Valoració de l'estereopsis per mitjà del test de Frisby. Ens permet examinar la fusió sensorial del pacient i valorar si suprimeix o no, i si fusiona o no.

Cap de les proves realitzades pot causar cap tipus de risc ni són invasives.

Per qualsevol dubte o problema pot posar-se en contacte amb Mireia Pacheco pacheco@oo.upc.edu o Elvira Peris peris@oo.upc.edu.

Annex II: Individual recorded sheet of the clinical exploration

DADES PERSONALS				
NOM		EDAT		
H. DEL CAS				
MEDICACIÓ				
HOBBIES				
RX HABIT	SI	FRONTO	UD	US:
	NO		UE	

EXAMEN REFRACTIU				
AV (3m)	UD		PPC:	
	UE			
CT VL		COMITÀNCIA		
CT VP				
AUTOREFRACTÒMETRE	UD			
	UE			
REFRACCIÓ	UD		AD	
	UE			
ST				

EXAMEN FUNCIÓ ACOMODATIVA		
NOT (20cm)	UD	
	UE	
NOTT	UD	
	UE	

SALUD OCULAR		
VISIÓ DEL COLOR		
FONS D'ULL	UD	
	UE	
OBSERVACIONS		