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Original article

Trochlear dysplasia and patellar instability in patients with Down syndrome[☆]



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ARTICLE INFO

Article history:

Received 23 August 2013

Accepted 7 March 2014

Available online 30 March 2015

Keywords:

Patellar dislocation

Down syndrome

Knee

ABSTRACT

Objective: To analyze occurrences of trochlear dysplasia in patients with Down syndrome in the presence and absence of femoropatellar instability.

Methods: Eleven knees with stable patellae and thirteen with unstable patellae in patients with Down syndrome were compared. Radiographs were produced to evaluate patellar height, trochlear angle and femoropatellar congruence angle.

Results: The prevalence ratio for a high patella between the unstable and the stable patients was 1.01 using the Insall-Salvati index and 0.68 using the Caton-Deschamps index. For an abnormal congruence angle, the prevalence ratio was 2.04. An increased congruence angle was only found in four cases, all presenting instability.

Conclusions: Trochlear dysplasia was only found in cases of instability. The trochlear groove angle and the femoropatellar congruence angle correlated with the presence of patellar instability.

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Displasia da tróclea e instabilidade patelar em pacientes com síndrome de Down

RESUMO

Objetivo: Analisar displasia troclear em pacientes portadores de síndrome de Down na presença e na ausência da instabilidade femoropatelar.

Métodos: Comparação de 11 joelhos com patelas estáveis e 13 joelhos com patelas instáveis em portadores de síndrome de Down. Foram feitas radiografias em que foram avaliados altura patelar, ângulo da tróclea e ângulo de congruência femoropatelar.

Palavras-chave:

Luxação patelar

Síndrome de Down

Joelho

[☆] Work developed in the Knee Group, Institute of Orthopedics and Traumatology, Hospital das Clínicas, School of Medicine, Universidade de São Paulo (HC/FMUSP), São Paulo, SP, Brazil.

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<http://dx.doi.org/10.1016/j.rboe.2015.03.005>

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Resultados: A razão de prevalência de patela alta entre os pacientes instáveis e os pacientes estáveis foi de 1,01 para o índice de Insall-Salvati e de 0,68 para o índice de Caton-Deschamps. Para o ângulo de congruência alterado, a razão de prevalência foi de 2,04. O ângulo de congruência aumentado foi encontrado apenas em quatro casos, todos com instabilidade.

Conclusões: A displasia da tróclea foi encontrada apenas em casos de instabilidade, o ângulo do sulco da tróclea e o ângulo de congruência femoropatelar se correlacionaram com a presença de instabilidade patelar.

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Introduction

Femoropatellar instability is a frequently occurring pathological condition and is linked with predisposing factors in most of the patients. Among these, dysplasia of the femoral trochlea and the height of the patella are considered to be the most important predisposing factors.¹

Uncertainties remain with regard to understanding the etiopathogenesis of these factors, especially dysplasia of the trochlea. There is doubt concerning whether the dysplasia is the cause or the consequence. Thus, it is asked whether a congenital abnormality would lead to femoral dysplasia characterized by a less deep trochlea, which would favor instability, or whether muscle abnormalities would cause the patella to have an abnormal course, thereby reducing the femoropatellar pressure and giving rise to incorrect stimulus during the anatomical development of the trochlea, such that it would become flatter, with consequent progression to instability.

To understand these factors better, they need to be studied and analyzed in patients with early femoropatellar instability, i.e. in children while they are growing, which makes it possible to observe the development of the femoral trochlea.

Among individuals with Down syndrome, there are patients with severe femoropatellar instability together with early dislocation of the patella.² This peculiarity of this group of patients makes it possible to analyze the development of the femoral trochlea concomitantly with the reduction in stimulation resulting from the presence of the patella in the femoropatellar joint, in situations of early dislocation of the patella. This makes it possible to compare this development with that of patients with Down syndrome in whom femoropatellar instability is not present. In addition to the development of the trochlea, the height of the patella can be studied. This factor relates to the quadriceps muscle, since it affects the degree of flexion of the knee to which the patella is jointed with the trochlea, and also influences the stimulation of development of greater or lesser trochlear depth.

The aim of this study was to analyze the depth of the trochlea and height of the patella (predisposing factors) in patients with Down syndrome with femoropatellar instability and compare these with the occurrence of these factors in patients with Down syndrome without femoropatellar instability.

Materials and methods

This study was approved by the Research Ethics Committee under no. 175/2010. All the patients received a free and informed consent statement.

We studied 12 patients with Down syndrome aged over 12 years who had never undergone any interventions in the knees.

We had six male and six female patients. Their mean age was 16.4 years, with a range from six to 36 years (Table 1).

We evaluated femoropatellar instability according to clinical and radiographic criteria. The radiographic measurements were made twice, with a one-week interval between them, by the same evaluator.

Clinically, we used the Dugdale and Renshaw classification system to determine whether instability was present.² This divides femoropatellar instability in patients with Down syndrome into five grades, such that grade I is stable and grades II and onwards are unstable, and the higher the grade is, the greater the severity of the instability is.

From this sample, we obtained 11 knees with stable patellae and 13 with unstable patellae.

We radiographically studied the knees individually in accordance with the radiological instability criteria. For all 24 knees, we used the frontal radiographic view and the absolute and axial lateral views in the Merchant position, flexed at 45°. ^{1,3,4}

The following were measured on all the radiographs on these patients: the height of the patella, in accordance with the method of Caton and Deschamps,^{5,6} the trochlear groove angle, in accordance with the method described by Merchant; and the patellar congruence angle. This last measurement was made between the line bisecting the trochlear groove angle and the line going from the vertex of the trochlear angle to the eminence or crest between the patellar facets (Fig. 1).

In evaluating the height of the patella on the lateral radiograph of the knee, we considered the patella to be high when the ratio between the distance from the anterior border of the tibia to the lower center of the joint face of the patella and the measurement of the joint surface was greater than 1.2 (Fig. 2).

In evaluating the trochlear groove angle, we considered the trochlea to be flat when the angle was greater than 150°.

In evaluating the patellar congruence angle, we considered that lateral subluxation greater than 11° or complete dislocation represented positive or altered values.

Table 1 – Anthropometric description of the sample, including means and standard deviations.

Number	Sex	Date of birth	Age (a)	Weight (kg)	Height (m)	BMI
1	M	01/10/95	14	52	1.54	21.9
2	F	7/25/00	9	26	1.26	16.4
3	M	5/25/99	10	31	1.4	15.8
4	M	5/9/00	9	44	1.32	25.2
5	M	7/10/92	17	90	1.6	35.2
6	F	4/11/89	21	52	1.52	22.5
7	F	4/8/99	11	45	1.48	20.6
8	M	7/7/87	22	59	1.53	25.2
9	F	9/19/87	22	65	1.4	33.2
10	M	4/29/74	36	74	1.73	24.7
11	F	11/3/03	6	23	1.12	18.3
12	F	5/5/90	20	68	1.4	34.7
Mean			16.4	52.4	1.44	24.5
Standard deviation			8.4	20.2	0.16	6.75
Minimum			6	23	1.12	15.8
Maximum			36	90	1.73	35.2

Results

From the clinical criteria according to the Dugdale classification, 11 knees were stable and 13 were unstable (grade II or higher).

Among the unstable knees, eight presented subluxated patellae, three presented reducible dislocated patellae and two had irreducible patellae.

From the radiographic evaluation, a high patella was identified in three knees. The trochlear groove was altered in four knees and the patellar congruence angle was altered in 13 knees (Fig. 3).

Table 2 presents the results and correlates the instability according to clinical and radiographic criteria.

The prevalence ratio is a measurement that makes it possible to estimate the strength of association between the exposure (in this case, the presence of instability) and the disease or abnormality studied (in this case, the radiographic alterations).

The prevalence ratio of high patellae between the unstable and stable patients was 0.68, using the Caton-Deschamps index. No prevalence ratio could be calculated for increased

trochlear groove angle, because this only occurred in the unstable patients. The prevalence ratio for the congruence angle was 2.04.

Trochlear dysplasia was only found in patients presenting instability (four knees).

Discussion

Down syndrome is one of the commonest disorders of genetic origin in humans. It affects between 1:1000 and 1:700 live births. It consists of trisomy of chromosome 21 and can also occur due to Robertsonian translocation.⁷

Orthopedic disorders are frequently observed in the spine (atlantoaxial instability and scoliosis), feet, hips (instability

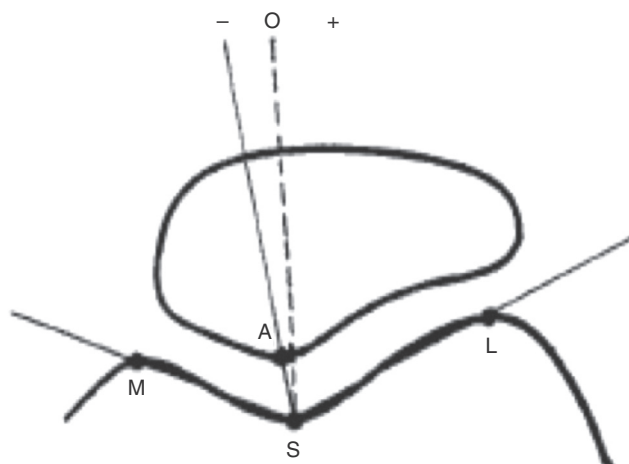


Fig. 1 – Diagram showing axial incidence on the knee, with measurement of the patellar congruence angle.



Fig. 2 – Measurement of the patellar height. It was considered high when $AT/P > 1.2$.

Table 2 – Correlation between instability according to clinical criteria and factors predisposing toward instability according to radiographic criteria.

Radiographic criterion	Clinical criterion	
	Stable	Unstable
High patella (Caton-Deschamps)	5	4
Trochlear groove >150°	0	4
Patellar congruence >11°	4	9

and sliding of the proximal femoral epiphysis) and knees (valgus knee and femoropatellar instability). These alterations are due to muscle disorders (hypotonia) and ligament laxity, which are common in this syndrome. Knee problems are seen in 30% to 40% of these patients.⁷

We consider that the age range of our patients was a limitation of our study, in which the mean was 16.4 years and the range was from six to 36 years. Studying the patients with Down syndrome is always difficult, especially when the individual is asymptomatic or has few symptoms of the pathological condition in question, given that the patients and their families are resistant to excessive medical attention. We studied 12 patients who had never had any medical attention for their orthopedic problems. Initially, we limited our inclusion criterion to patients aged under 14 years, but this was seen to be insufficient for obtaining an adequate sample. Therefore, we decided to also include patients over the age of 14 years.

Furthermore, we only used simple radiographs in our evaluation, because the patients had never undergone tomography, although the latter is considered to be the best examination for estimating the Q angle (by means of TA-GT) and rotational displacement of the limb. Since many patients presented high fat distribution to the thighs, we chose not to include a clinical measurement of the Q angle because of the low reliability of this analysis.

In order to define the presence of instability, we used the Dugdale and Renshaw classification,² because this is classical for patients with Down syndrome presenting femoropatellar instability. In this manner, we selected 13 knees with femoropatellar instability that had been diagnosed clinically

in accordance with the Dugdale criteria and we compared these with 11 knees that according to the same methodology did not present any femoropatellar instability, even though they were the knees of patients with Down syndrome. We chose to categorize our data analysis between knees with and without instability, since this analysis is more reliable than a classification of instability into several grades and also strengthens the data analysis in a sample of more restricted size. The radiographic criteria were chosen and limited to three positions. The three indices evaluated were based on the study by Dejour et al.,¹ with the aim of establishing the presence of objective signs of femoropatellar instability. During the study, we attempted to evaluate the depth of the trochlea on radiographs in absolute lateral view, but we only considered these measurements to be adequate in three patients and preferred to disregard this measurement of instability in the present study. Individuals with Down syndrome often do not cooperate during examinations and several radiographs may need to be produced in order to obtain one that is good enough for analysis. Increasing the number of radiographic views significantly complicates the study and exposes the patients to additional radiation. With regard to complementary examinations, we chose not to perform computed tomography because of the radiation exposure, given that these patients are frequently exposed to multiple examinations over their lifetimes. We chose not to perform magnetic resonance examinations because a large proportion of the patients would need to be sedated.

In our results, we observed that the patellar height (a femoropatellar instability index that is considered to be very important among patients without Down syndrome) was not useful as a criterion for instability here, given that among the nine knees with abnormal patellar height, five were stable. The patellar height evaluation method using the joint surface (Caton-Deschamps) was chosen because this presents results that are more reliable and have better correlation between the evaluators.⁸ Initially, we imagined that this might have been a problem consequent to the measurement method chosen (Caton-Deschamps) and therefore we repeated the patellar height measurements using the Insall-Salvati method.⁹ In this second measurement, we found that 22 knees presented a patella that was considered high, and ten were in patients in the group that was considered to be stable. This corroborated the finding that the patellar height is not an appropriate measurement for indicating femoropatellar instability in patients with Down syndrome. The prevalence ratio for high patella between the unstable and stable patients was only 1.01 according to the Insall-Salvati index.

Barnett et al.¹⁰ analyzed patellar height in magnetic resonance imaging examinations on completely extended knees, among 29 patients with trochlear dysplasia in association with patellar instability. They observed that there was no relationship between high patella and instability. We believe that muscle alterations relating to Down syndrome may be responsible for retraction of the quadriceps, which would cause a high patella in most patients, even in individuals who do not present femoropatellar instability.

The patellar congruence angle was positive for instability in nine of the 13 unstable knees and in four of the stable knees, which demonstrates that this measurement presents

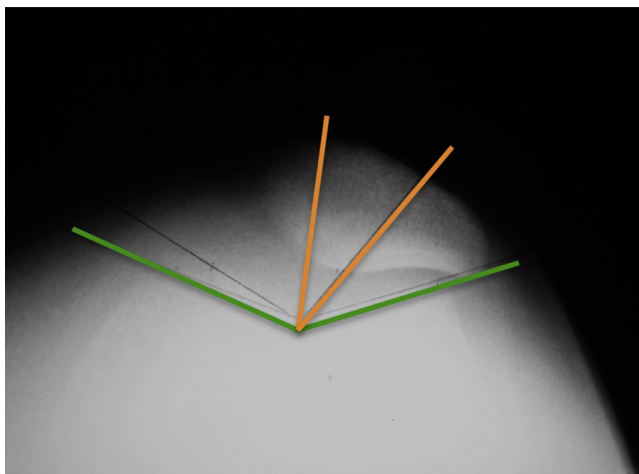


Fig. 3 – Radiograph of the knee in axial view. Note the trochlear dysplasia and lateralization of the patella.

a higher prevalence ratio (2.04) in situations of femoropatellar instability.

The measurements defining the depth of the trochlea were clearly positive for femoropatellar instability in the patients whose patellae had become dislocated and subluxated. Among the four knees with alterations of the femoral trochlear angle, all of them were unstable.

The prevalence ratio calculations in the present study demonstrated that the prevalence of altered congruence angles in patients with patellar instability was 2.04 times greater than in stable patients. The prevalence ratio is a more conservative measurement than the odds ratio.¹¹ Since this was a cross-sectional study and not a prospective case-control study, we chose to use prevalence ratios. Odds ratio calculations on our sample would result in values of 1.2 and 0.53 for the patellar height according to the Insall-Salvati and Caton-Deschamps indices and 6.75 for alterations of the congruence angle. These data reinforce our understanding that the height of the patella is not a reliable index for assessing patellar instability among patients with Down syndrome, whereas the patellar congruence angle is relevant among these patients, as is the groove angle, which was found to be altered only in unstable knees.

We believe that absence of the patella in the femoral trochlea impeded formation of the trochlear groove. This is suggested from the observation that patients without instability who presented Down syndromes had normal trochlear depths. Studies using magnetic resonance imaging have demonstrated that patients with non-traumatic patellar instability have high prevalence of trochleae with lateral condyle inclinations less than 11°. ^{12,13} In our view, this means that the trochlea is shallower when there is less demand for the presence of the patella, thereby giving rise to larger angles. The presence of the patella aligned on the trochlear groove is what determines its depth and normal shape.

Dejour et al.¹⁴ believed that trochlear dysplasia was a congenital alteration that would be a predisposing factor favoring femoropatellar instability. If this were so, trochlear dysplasia might be found in young patients who had not developed instability, which was not seen in the sample studied here. In our view, trochlear dysplasia cannot be taken to be congenital, at least in patients with Down syndrome.

Finally, it needs to be borne in mind that the osteoarticular alterations seen in Down syndrome are generally multiple and that care is required in transposing these observations to patients without Down syndrome.

Conclusion

From the sample studied, the findings suggest that trochlear dysplasia is not congenital, but develops as a result of patellar instability.

The radiographic measurements on the femoral trochlear depth showed that the trochlea was at more than 150° (flat) only in cases of instability.

Basic radiographic measurements, such as measurements of the trochlear groove angle, correlated with the femoropatellar instability.

Conflicts of interest

The authors declare no conflicts of interest.

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