European Journal of Pediatrics (2019) 178:559–564
https://doi.org/10.1007/s00431-019-03322-x

ORIGINAL ARTICLE

Developmental dysplasia of the hip in children with Down syndrome: comparison of clinical and radiological examinations in a local cohort

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Received: 24 August 2018 / Revised: 27 December 2018 / Accepted: 10 January 2019 / Published online: 1 February 2019

Abstract

Guidelines for children with Down syndrome (DS) suggest to perform an annual hip screening to enable early detection of developmental dysplasia of the hip (DDH). How to perform this screening is not described. Delayed detection can result in disabling osteoarthritis of the hip. Therefore, we determined the association between clinical history, physical, and radiological examination in diagnosing DDH in children with DS. Referral centers for children with DS were interviewed to explore variety of hip examination throughout the Netherlands. Clinical features of 96 outclinic children were retrospectively collected. Clinical history was taken, physical examination was performed, and X-ray of the hip was analyzed. All the referral centers performed physical examination and clinical history; however, 20% performed X-ray. Following physical examination according to Galeazzi test 26.9% and to limited abduction 10.8% of the outclinic-studied children were at risk for DDH. Radiological examination showed moderate or severe abnormal deviating migration rate of 14.6% resp. 11.5% in the right and left hip. However, no association between clinical history, physical examination, and radiological examination was found.

Conclusion: Clinical history and physical examination are insufficient to timely detect DDH in children with Down syndrome. Thereby regular radiological examination of the hip is advised.

What is Known:
- *Developmental dysplasia of the hip (DDH) in people with Down syndrome (DS) develops during childhood.*
- *Guidelines for medical support of children with DS suggest an annual hip screening to enable early detection of hip damaging. How to perform this annual screening is not described.*

What is New:
- *This study shows no association between clinical history, physical and radiological examination of the hip.*
- *We recommend regular radiological examination of the hip in children with DS in order to identify DDH early up to 16 years of age.*

Keywords Developmental dysplasia of the hip · Down syndrome · Multidisciplinary guidelines · Radiological examination

Communicated by Mario Bianchetti

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Abbreviations

DDH	Developmental dysplasia of the hip
DS	Down Syndrome
L	Left hip
MMC	Máxima Medical Center
R	Right hip
SD	Standard deviation

Introduction

Down syndrome is a common chromosomal abnormality, with a birth incidence of 1 per 741 in the Netherlands [1]. The age of mothers at the first birth is increasing. These mothers have a statistically higher risk for chromosome abnormalities and thereby the incidence of DS can increase [2]. Developmental dysplasia of the hip (DDH) is fairly common in adults with DS, with an estimated incidence ranging from 1.25–7%, with higher numbers recorded among institutionalized patients [3, 4]. Due to improved medical care, life expectancy in Down syndrome has increased significantly in the past decades, from 12 years in 1940s to average 60 years nowadays in developed countries [5]. Thereby, the prevention of orthopedic problems becomes more important.

Developmental dysplasia of the hip in persons with DS already develops in childhood. The development of hip instability can be classified into four phases: initial phase (0–2 years of age), dislocation phase (2–8 years of age), subluxation phase (> 8 years of age), and fixed phase (> 15 years of age) [6, 7]. Although irrespective of the age, a hip can always start to dislocate, in particular children with DS. The initial phase is characterized in newborns with a reducible dislocation of the hip which are expectantly treated. In the subluxation phase children already develop secondary acetabular pathophysiology, for example developmental dysplasia of the hip (DDH) with and without subluxation. The intermediate phase, the dislocation phase, is characterized by marked soft tissue laxity without secondary acetabular pathophysiology. Therefore it is important to detect DDH around the dislocation phase where there is no secondary pathophysiology and therefore treatment is less complex [6]. This contributes to maintaining mobility, and reducing healthcare costs.

Searching worldwide two guidelines are published in the Netherlands and the USA for medical support of children with DS [1, 8]. These guidelines advise an annual hip screening to enable early detection of hip damaging in children with Down syndrome. However, how to perform this annual hip screening is not described in these guidelines. In order to determine the best way to timely detect developmental dysplasia of the hip in children with Down syndrome, we studied clinical history, physical examination, and radiological examination, and their interrelationship.

However, sensitivity and specificity of the usual physical examination of the hip were determined on studies in newborns with congenital hip dysplasia. Congenital hip dysplasia has a different pathological mechanism than developmental dysplasia of the hip in Down syndrome [9]. There is no evidence that newborns with Down syndrome have an increased risk of congenital hip dislocation, or have an increased risk of an unstable hip joint as a newborn [10]. Physical examination of the hip has not been studied in children with Down syndrome.

Methods

Interview with referral centers

The pediatrician referral centers for children with Down syndrome in the Netherlands ($n = 15$) were interviewed to explore the variety in performing clinical history, physical examination, and radiological examination as advised by the Dutch guideline.

Study design and population

This study was conducted at Máxima Medical Center (MMC) in Veldhoven, The Netherlands. MMC is a referral center for children with Down syndrome in the South-East of the Netherlands. A retrospective study using hospital medical records was conducted. Walking children aged 2–18 years with Down syndrome visiting the outpatient Down clinic on a regular annual basis at the Máxima Medical Center, with an X-ray of the hip in the system, were included ($n = 96$). Children from other referral areas were excluded. All children had chromosomal regular trisomy 21. None of the children were wheelchair bound, one hemiplegic child was excluded. Children with hip pathology not due to developmental dysplasia of the hip as Perthes disease or epiphysiolysis ($n = 2$) were excluded.

Clinical history and physical examination

The studied data for clinical history were family history of hip dysplasia and clinical history of hip pain. Physical examination was performed by a child physical therapist, examining gait, audible or palpable inducible click, inducible hip dislocation, abduction test, Galeazzi test, and degree of laxity using Bulbenascore [11]. The data for clinical history and physical examination were searched before the age of the child when the X-ray of the hip was made.

Radiological examination

Radiological examination was studied in standing position with an X-ray of the hip. All radiological examinations were independently assessed by two of the authors (ER and FvD). In case of discrepancies, consensus was reached between the two observers.

Studied items in the radiological examinations were up till 8 years of age the acetabular angle as measured by Sharp, as this angle is only reliable for the children up to 8 years of age, with normal values differ per age [12, 13]. The center-edge angle of Wiberg, which is reliable for children above 8 years of age, was classified as normal if > 25°, mild aberrant if between 20° and 25° and severely aberrant if < 20° [13–15]. Migration percentage was measured and radiographic hip subluxation of the head or the hip was defined as mild (10–20%), moderate (20–30%), or severe (> 30%) uncovered.

Developmental dysplasia of the hip with or without luxation was defined as either painful or limited walking, dislocation at physical examination, severe abnormal acetabular angle > 30°or center-edge angle < 20°, or migration percentage of more than 30%.

Statistical analysis

Descriptive statistics were used to describe the distribution of primary outcome and covariates. The prevalence was expressed as the percentage of patients with DDH. Associations between radiological findings and physical examination or clinical history findings were analyzed by using univariate logistic regression analysis. In this analysis, we included migration percentage as the dependent variable. Odd ratio and confidence intervals are reported as OR and CI. Statistical significance was accepted at a two-sided p value < 0.05. Statistical analyses were performed using the SPSS 19 software.

This study was approved by the accredited medical ethical review board of the MMC.

Results

Interview with referral centers

The pediatrician referral centers for children with Down syndrome in the Netherlands were interviewed ($n = 15$). All referral centers performed physical examination and took clinical history of the hip by the pediatrician on an annual basis. Standard radiological examination performing an X-ray of the hips every 2 years was performed in three of the referral centers (20%). The other 80% performed radiological examination of the hip if indicated by physical examination or clinical history.

Baseline characteristics

In this study, 96 children were included, 58 male (60%) and 38 female (40%). The mean age of the study population was 6.4 years (SD 4.3 years).

Clinical history and physical examination

Characteristics of clinical history and physical examination are shown in Table 1. The most prevalent findings were hyperlaxity of the hip ($n = 20$, 21.3%) and deviating Galeazzi test ($n = 25$, 26.9%). Walking remarks ($n = 8$) that were noted included: walking with circumduction, more exorotation in one leg and walking with an unequal stride length.

Radiological examination

The results of the radiological examination are shown in Table 2. Abnormalities in acetabular and center-edge angle were rare, but abnormal deviating migration was highly prevalent. Moderate to severe abnormal deviation of the right hip was seen in 14 hips (14.6%), 8 boys (13.8%) and 6 girls (15.8%), and severe abnormal deviation of the left hips was seen in 11 hips (11.5%), 6 boys (10.3%) and 5 girls (13.1%); one boy had abnormal deviation in left and right hip.

Logistic regression analysis

No significant associations were found between radiological examination and physical examination, nor between radiological examination and gender. Logistic regression analysis showed no significant association between migration percentage and increased laxity both for the right and left hip (OR 2.407, CI 0.704 to 8.228, $p = 0.161$ and OR 0.802, CI 0.159 to 4.050, $p = 0.790$. Also, no association was found between complaints of pain, physical, and radiological examination. There was no significant association between migration rate

Table 1 Results of clinical history and physical examination

Clinical history ($n = 96$)	Missing value	Negative	Positive
Hip dysplasia in family	0	92 (95.8%)	4
Hip pain	2	94 (100%)	0
Physical examination			
Hyperlaxity**	2	74 (78.7%)	20 (R + L)
Click audible or perceptible	2	94 (100%)	0
Hip luxation provoke	2	93 (98.9%)	1 (R)
Walking problems	2 + 8*	78 (83%)	8
Abduction limitation (R/L)**	3	83 (89.2%)	4/6
Galeazzi deviating (R/L)**	3	68 (73.1%)	16/9

*Cannot walk yet; **R = right hip; L = left hip

Table 2 Results of radiological hip examination

	Missing value (R/L)**	Number (R/L)**	Normal (R/L)**	Mild (R/L)**	Moderate (R/L)**	Severe (R/L)**
Acetabular angle deviating (age ≤ 8 y)*	0/1	63/62	62/58	1/3	0/1	0/0
Center-edge angle deviating (age ≥ 9 y)*	0/0	33/33	32/32	1/1	0/0	0/0
Migration percentage deviating***	0/0	96/96	50/72	32/13	10/9	4/2

*y = age in years; **R = right hip; L = left hip; *** Migration percentage defined as normal < 10%, mild abnormal 10–20%, moderate abnormal 20–30%, or severe abnormal ≥ 30%

and abduction limitation both for the right and left hip (OR 1.795, CI 0.173 to 18.620, $p = 0.624$ and OR 4.687, CI 0.739 to 29.731, $p = 0.101$). Also, there were no significant relations between migration rate and Galeazzi test both for the right and left hip (OR 0.345, CI 0.41 to 2.891, $p = 0.327$ and OR 2.490, CI 0.430 to 14.4.05, $p = 0.308$).

Discussion

Developmental dysplasia of the hip in persons with Down syndrome already develops in childhood. Thereby, the Dutch national multidisciplinary guideline for medical support of children with Down syndrome advise an annual hip screening [1]. However, this guideline does not describe how to perform screening of the hip by performing a clinical history, physical, and/or radiological examination. Concluding out of the interview with the referral centers for children with Down syndrome in the Netherlands, only 20% performed a radiological examination by performing an X-ray of the hips every 2 years. The other 80% performed radiological examination of the hip only if indicated by physical examination or clinical history. Therefore, the risk to detect developmental dysplasia of the hip in an advanced stage is probably high and possibly to late [6]. As described by Knight et al. management of developmental dysplasia of the hip with a subluxation or dislocation in children with DS includes surgery [16, 17]. They suggest to perform femoral varus derotation osteotomy before the age of 7 years old for a predictable result [16].

In order to timely detect DDH, literature advises to use ultrasound [18]. But, in the Netherlands, we are still used to perform X-ray of the hips after the neonatal period, also to evaluate the migration percentages. As migration index is calculated using X-ray.

There is little reported in literature about children with Down syndrome as to whether clinical history and physical examination can be related to radiological findings. In our study, no association was found between clinical history, physical, and radiological examination. Results of physical examination were not associated with radiological examination.

In this study, the presence or absence of pain at clinical history or physical examination does not discriminate between the presence or absence of DDH in children with Down syndrome. Valkenburg et al. describe that only a minority of children with Down syndrome are able to reliably self-report pain [15]. The data in this study suggest that clinical history nor physical examination can be used as a predictive modality to diagnose developmental dysplasia of the hip in children with Down syndrome and therefore we advise regular radiological examination of the hip (Figs. 1 and 2).

A remarkable finding in family history in this study was that out of four children with a positive family history for congenital hip dysplasia, two children had a severe abnormal migration percentage. It is unknown whether the occurrence of congenital hip dysplasia in family history is a risk factor in children with Down syndrome for developmental dysplasia of the hip in later life. These data suggest that this may play a role.

Incidence of Down syndrome is equal in boys and girls. In non-Down children congenital hip dysplasia is seen more often in girls than in boys (4:1); whether this is similar for Down syndrome is unknown. In this cohort, surprisingly 4 of 5 children with serious abnormal migration were boys (one boy with L and R severe deviation). This can only partially be explained that there were more boys than girls in this study

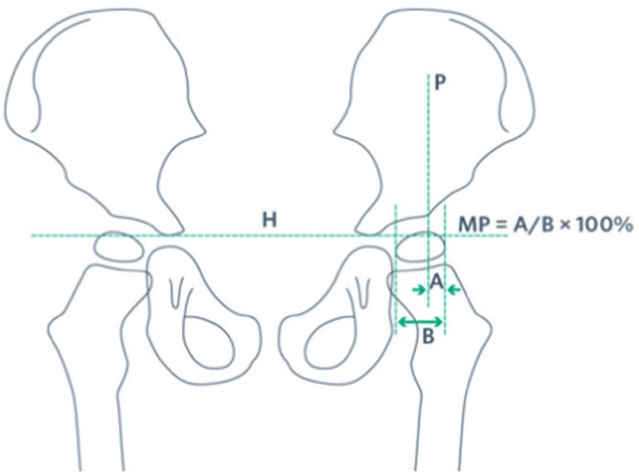

Fig. 1 Migration percentage is calculated by dividing width of uncovered femoral head (A) by total width of femoral head (B)

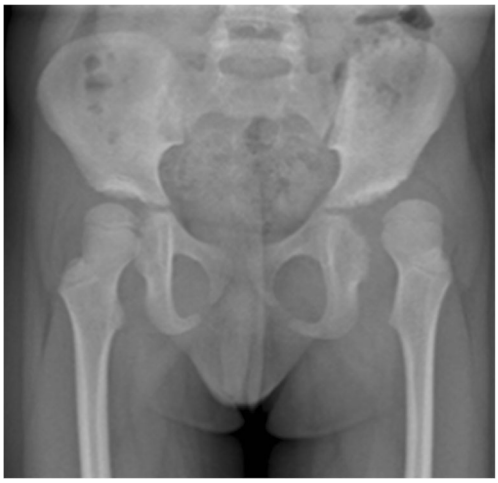

Fig. 2 X-ray of the hip in a 7-year old boy with DS. Migration percentage of the left hip (right side in the image) of 59%

population (60.4% versus 39.6%). There is no selection bias for hip abnormalities in this outpatient clinic. So no good explanation was found for the surplus of boys with severe hip dysplasia. Further studies have to focus on gender and hip dysplasia in children with Down syndrome.

It remains unclear why children with Down syndrome are at risk to develop hip dysplasia. Incidence of congenital hip dysplasia in the general population is 0.5%. In these children, the skeletal part of the hip is not well developed with an abnormal acetabular angle and abnormal center-edge angle. Although in children with Down syndrome there are slight variations in the construction of the acetabulum, such as a deficiency of the posterior acetabular wall [17]. Although this is insufficient to explain the instability of the hip in children with Down syndrome [19]. It has been described that there is an increase in acetabular depth, a decrease in acetabular anteversion and a roof which is more horizontal than normal [20]. A hip joint in a normal child has a high intrinsic stability through the osseous relations, the negative pressure in the joint and the very strong ligamentous structures complemented by an extensive and well-developed muscle cuff. In this cohort, the majority of children had a normal skeleton with a normal acetabulum, a normal acetabular angle and center-edge angle along with an abnormal migration percentage. This suggests that the pathogenesis of hip abnormalities in Down syndrome is different from children with congenital hip dysplasia, as the patients with severe deviating migration percentage had a normally developed hip skeleton.

Earlier publications mentioned that an increased laxity of the ligaments is a possible cause of the development of hip dysplasia in children with Down syndrome [6, 21]. In children without clinical signs of increased laxity, a deviating migration percentage was seen in 12.2% of the hips on radiological examination (in both left and right hip). In children with clinical signs of increased laxity a deviating migration percentage was seen in 10 vs 20% in respectively left and right hip. Absence of laxity did not exclude a possible hip abnormality nor did increased laxity indicate radiological hip abnormalities in this study. Only one earlier publication mentioned that laxity is not a major etiological factor for joint problems in children with DS as well [22].

Advantages of this study compared to previous studies is that this studied population is a large unselected group, all children were between the age of 2–18 years, no one was wheelchair bound and little data were missing.

Conclusion

Clinical history taking and physical examination are insufficient to determine developmental dysplasia of the hip in children with Down syndrome. DDH on radiological examination is highly prevalent and shows abnormal findings in ($n = 14$, 14.6% and $n = 11$, 11.5% in the right and left hip respectively) of children with Down syndrome. Multidisciplinary guidelines for medical support of children with Down syndrome advise to perform an annual hip screening; however, no suggestion which examination to perform is made. Interpretation of the guideline by different persons results in different way of investigation. In the Netherlands, only 20 % of the referral centers for children with Down syndrome perform an X-ray of the hip on a regular basis, this leads to a risk to detect DDH in a more severe stage then needed. We recommend that guidelines in Down syndrome should advise radiological examination of the hip in children with Down syndrome up to at least 16 years of age in order to identify DDH early. Early detection, referral, and treatment should be aimed to improve mobility and improved long-term outcome.

Authors' contributions AFM van Gijzen has had a substantial contribution to the analysis and interpretation of the study. She added substantial data and results to the paper and rewrote the paper and processed the giving feedback. Agree to be accountable for all aspects of the work.

As a student, EDM Rouers has collected all the data and the analysis and wrote the first draft of the paper. Agree to be accountable for all aspects of the work.

QMP van Douveren has delivered a substantial contribution to the design of the study and acquisition of the data. Agree to be accountable for all aspects of the work.

J Dieleman had a great part in the study design and the data analysis. Agree to be accountable for all aspects of the work.

GE Hendriks has delivered a substantial contribution to the design and analysis of the study and acquisition of the data. Agree to be accountable for all aspects of the work.

F Halbertsma has had a substantial contribution to the design of the study, analysis, and interpretation of the study. Agree to be accountable for all aspects of the work.

LA Bok has had a substantial contribution to the design of the study, the acquisition, analysis, and interpretation. Agree to be accountable for all aspects of the work.

Compliance with ethical standards

This study was approved by the Medical Ethical Commission. All procedures performed in studies were done according to regular yearly medical care for children with Down syndrome, studied in retrospect, and in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Competing interests The authors declare that they have no competing interests.

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