Original

Predictive Factors for the Development of Vertebral Deformities in Down's Syndrome

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Abstract

Background: There are many studies of spine pathology in people with Down Syndrome (DS), the majority of these focusing on atlantoaxial anomalies of the cervical column. There is, in contrast, much less information about other malformations such as scoliosis, kyphosis and lordosis, perhaps due to the fact that they are much less frequent pathologies with few physical and life-threatening repercussions. The aim of our study was to determine the prevalence of these spine-related malformations in a group of patients with DS and to look for factors that might predict the course of their development.

Patients and methods: We studied a total of 60 people with DS, 26 women (43.33) and 34 men (56.67%) with an average age of 39.8 ± 10.19 years, weight average of 65.4 ± 13.55 kg, and height average of 1.51 ± 0.4 metres. In all cases, we studied the different alterations of the spine following a well-established three-dimensional study protocol. We studied anthropometric data for these patients and also evaluated the other alterations which, due to their frequency, were thought to be potentially related to other spine-induced malformations: thyroidal pathologies, heart- and sight-related problems and alterations affecting the locomotive apparatus such as atlantoaxial instability, hyperlaxitude of the ligaments and deformation of the lower limbs.

Results: Forty-nine (81.67 %) of the patients had some spine-related malformation, 21 (35%) had malformations related with scoliosis, 19 (31.67%) with kyphosis, 30 (50%) with lumbar hyperlordosis, and 15 (25%) suffered from 2 or more of these malformations. Sex and height did not seem to be predictive factors, but age seemed related to lumbar lordosis, with this condition being more prevalent in younger patients ($p < 0.01$), while dorsal kyphosis was more prevalent in older patients ($p < 0.001$). On crossing spine-related malformations with other disorders, we noted that patients who suffered some form of visual pathology had a higher risk of suffering from scoliosis ($p < 0.0009$) and also other types of skeletal malformation ($p < 0.02$).

Conclusions: The number of patients with spine-related malformations was very high in the DS study group. With time, the curvatures change: in young people bone curvature predominantly affects the lumbar hyperlordosis, while in older patients it affects the dorsal kyphosis. Patients with visual disorders and other malformations of the skeleton have a greater risk of suffering scoliosis. We think that in this population group it is very important to prevent or to reduce the column deviations.

Key words: Down's syndrome. Risk factors. Vertebral deformities.

Introduction

Down syndrome (DS) results from chromosome 21 trisomy and is one of the most common genetic abnormalities. Individuals with this syndrome have various associated medical conditions. Orthopedic conditions are common, and there have been reports of a relationship between DS and joint and ligamentous laxity, atlantoaxial instability, and abnormalities in the morphology of the foot, among others (1-3). However, there are few studies that discuss other spinal deformities such as scoliosis, kyphosis, or hyperlordosis (4,5), perhaps because these alterations have little physical or life-threatening impact. The onset of these curvatures of the spine suggests the possible presence of a cause that promotes the condition, such as age, weight, laxity, or association with other conditions described as enhancing idiopathic scoliosis in adulthood, for instance thyroid, visual, or cardiac conditions or deformities of the lower limbs, in particular, the feet (6-8).
The purpose of our study was to determine the prevalence of these spinal deformities in a group of persons with DS and to identify any predictive factors related to their development.

Materials and Methods

Sixty subjects with DS were studied, all employed at two special work centers. In all cases, the patient was older than 20 years at the time of the first examination. Individuals with severe mental impairment were excluded from the study.

Overall Examination

The age, height, and weight were recorded and an assessment was performed on the presence of various conditions potentially associated with static abnormalities, such as visual or cardiac conditions, hypothyroidism, and other skeletal deformities, including atlantoaxial instability, ligamentous laxity, or deformities of the lower limbs such as flat, high-arch or pronated feet.

Specific Clinical Examination

1. In order to study the various deformities of the spine, a protocol recently described by C. Marty (9) was followed with regards to definitions, clinical examination, and diagnosis. The studies were conducted by observing the three spatial planes.

In the frontal plane, a plumb line was used to measure from the C7 apophysis downward and a ruler was used to measure the distance between the plumb line and the curvature. Any lateral asymmetry or difference in shoulder height was recorded (Figure 1).

In the sagittal plane, a plumb line and goniometer were used. The different forms were recorded: normal, curvature inversion, dorsal or lumbar kyphosis, and flat back.

In the horizontal plane, the patients were examined standing and seated. The presence of any hunchbacks that would differentiate between true scoliosis and a scoliotic posture was recorded.

2. Waist and hip examination: any abnormalities in hip and hamstring mobility were recorded, as well as any retraction of the pectoral muscles. Feet were examined and the various malformations of the arch recorded.

3. Neurological examination: the musculature, reflexes, sensitivity, and cranial nerves were assessed, and any intake of medication that could cause static disorders of the spine were taken into account.

4. To identify any correlation with other conditions such as visual, cardiac, or thyroid disturbances, the patient history kept in the medical records for each workshop was reviewed.

Radiological Examination

Plain front and side x-rays were requested for both diagnostic and follow-up purposes, as well as scoliosis x-rays in questionable cases. Two cases are shown: 1 with scoliosis (Figure 2) and 1 with flat back and hyperlordosis (Figure 3).

Statistical Analysis

This was a prospective, observational study in a high-risk population consisting of subjects with DS, which assessed the association between different risk factors and the appearance of spinal deformities. All variables are dichotomous except for age, height, and weight. All quantitative variables are expressed as arithmetic mean with standard deviation, with others expressed as relative frequencies or percentages. The quantitative variables were compared using

Figure 1. Goniometer, hammer, plumb line.

Student's t-test and analysis of variance (ANOVA), and the qualitative variables using the chi-square test. The test was considered to be statistically significant when $P<.05$, for a 95% confidence interval. The statistical analysis was done using SPSS 12.0 and G-Stat 2.1

Results

A total of 60 individuals with DS were studied, 34 men (56.67%) and 26 women (43.33%). The mean age was 39.8±10.19 years, mean weight, 65.4±13.55 kg, and mean height, 1.51±0.4 m.

A total of 49 individuals (81.67%) had some spinal deformity, 21 (35%) had scoliosis, 19 (31.67%), kyphosis, and 30 (50%), hyperlordosis. Fifteen individuals (25%) had more than 1 of these malformations.

We found no significant gender-related differences among the various parameters studied.

No statistical correlation was found between overall spinal deformities and the different variables. Conversely, we independently observed that individuals with some type of visual disturbance were at greater risk of scoliosis ($P<.0009$), and that persons who present skeletal malformations other than spinal deformities are at also at greater risk of this condition ($P<.02$).

Among the youngest, there was higher prevalence of lordosis ($P<.01$) and among the oldest, of kyphosis ($P<.01$).

None of the other study parameters (weight, height, ligamentous laxity, presence of thyroid or cardiac abnormalities) could be independently correlated to any of the spinal deformities.

Figure 2. Dorsolumbar scoliosis.

Figure 3. Flat back and hyperlordosis.

Discussion

Static disorders of the spine are a little-known field that has been barely studied. On most occasions, pediatricians or pediatric orthopedists are responsible for discovering any disorders of this kind. However, the problems resulting from spinal static deformities may persist for the entire life of affected patients. According to the degree of involvement, the patient may have vertebral pain or backache with compression-induced nerve or spinal cord complications, respiratory distress, or simply secondary esthetic or psychological alterations (10). In addition, these static abnormalities of the spine occur in persons who already have other congenital, muscle, or bone malformations that may enhance the problems. Patients with genetic diseases such as Marfan syndrome may have differing curvatures of the spine (11), a manifestation also seen in Prader-Willi syndrome (12) or Charcot-Marie-Tooth disease (13), among others.

The factors involved in the development of various spinal deformities have still not been identified, and the hypotheses are countless and often contradictory. In a recent article (14), JL Tassin described research on the etiology and mentioned genetic factors—often extremely evident, since cases were found in the same family—, although the type of transmission was unknown. He also mentioned factors related to delayed maturation of balance or abnormalities in the biochemical constitution of intervertebral discs, among other more improbable theories.

DS is also a genetic syndrome caused by the presence of a third chromosome 21, which produces an extremely varied phenotypic expression with a higher prevalence of endocrine, visual, cardiac, and orthopedic disturbances with a predominance of hypotonia, joint laxity, and atlantoaxial instability with secondary dislocation (1-4). However, other spinal deformities have been studied very little and, therefore, we decided to undertake this study, in which we observed that 81.67% of the patients studied had some type of spinal malformation, with hyperlordosis and flat back the most common (50%), particularly between younger patients, that nevertheless led over the years to dorsal kyphosis. We also observed that individuals with some kind of visual disturbance leading to loss of vision were more likely to have scoliosis, and that those with some kind of skeletal deformity, such as flat, high-arch or pronated feet, had associated spinal deformities. Although we did not study the clinical repercussions of these malformations—basically due to the difficulties involved in questioning many of these individuals—, we feel that, as mentioned earlier, these are additional problems to those derived from DS.

At the time of the study, all subjects were involved in either swimming or physical therapy of various kinds, usually in a group setting, although some of the more difficult patients had personalized treatment.

Table I.
Study Parameters According to Gender

Parameters	Men	Women	P
Age (%)	41.08 (9.2)	38.1 (11.3)	NS
Weight (SD) (kg).	65.38 (10.9)	64.26 (16.5)	NS
Height (SD) (m)	1.55 (0.07)	1.44 (0.08)	NS
Spinal deformities. (%)	79.41	84.62	NS
Scoliosis (%)	41.18	26.92	NS
Kyphosis (%)	29.41	34.62	NS
Lordosis (%)	47.06	53.85	NS
Laxity (%)	23.53	34.62	NS
Thyroid abnormal. (%)	20.59	11.54	NS
Visual disturban. (%)	47.06	57.69	NS
Cardiac abnormal. (%)	29.41	34.62	NS
Other deformities (%)	58.82	78.92	NS

Data expressed as mean and standard deviation. NS: insignificant.

Conclusions

1. The prevalence of spinal deformities in the group of individuals with DS studied was very high and, unfortunately, little known.
2. Curvature gradually changes with age: hyperlordosis predominates among the youngest and dorsal kyphosis among the oldest. Individuals who have visual disorders and other skeletal deformities are at greater risk of scoliosis.
3. Its consequences on functional prognosis as well as the prognosis and quality of life should lead to the development of therapeutic and research programs to prevent or reduce the onset of spinal deviations that can complicate the progress of patients with DS.

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