Down's Syndrome Presented with Clubfoot Deformity : A Case Report

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Abstract

A 1 month old girl was referred to the orthopaedic clinic with bilateral clubfoot deformities. At birth, clinical examination showed the typical characters of Down's syndrome and the diagnosis was confirmed by chromosome study. These two conditions appear improbable as their basic pathologies are entirely different. In our patient, the translocation type at the long arm of chromosome 21 was determined in the chromosome study. This result has never been reported in the literature.

Key word : Down's Syndrome, Clubfoot, Chromosome Study

Down's syndrome is the most common autosomal chromosome abnormality. The orthopaedic problems involve mainly the consequence of the hyperlaxity including atlantoaxial instability, dislocation of the patella, spontaneous habitual dislocation of the hip, genu valgus, severe flexible pes planus and metatarsus adductus[1-6]. In contrast, the basic pathology of clubfoot is the contracture of the soft tissue, tendon and ligament surrounding the ankle, subtalar and mid-tarsal joint.

We report a patient with Down's syndrome who presented with bilateral clubfoot deformities.

CASE REPORT

A 1-month-old girl with Down's syndrome was referred to the pediatric orthopaedic clinic for treatment of bilateral clubfoot deformities.

She was the first child of a 29-year-old father and a 25-year-old mother. Both of them were office workers. The gestational period was 38 weeks. She was delivered by cesarean section due to labor pain and cephalopelvic disproportion. The body weight was 2700 grams. The familial history was negative for the occurrence of all congenital abnormalities.

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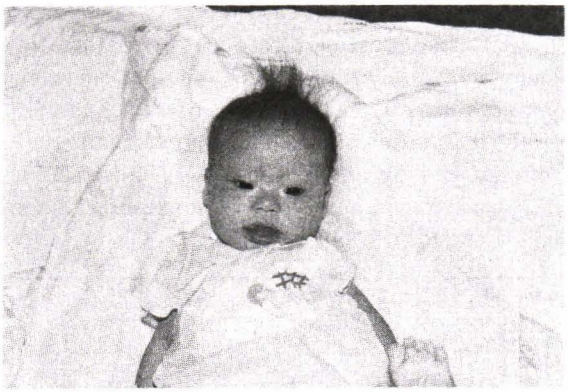
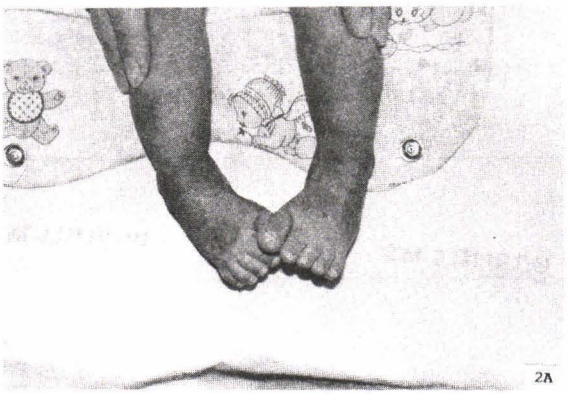


Fig. 1. Showing the typical findings of Down's syndrome such as microbrachycephaly, up-slanted eyes with epicanthal folds, protruding tongue.



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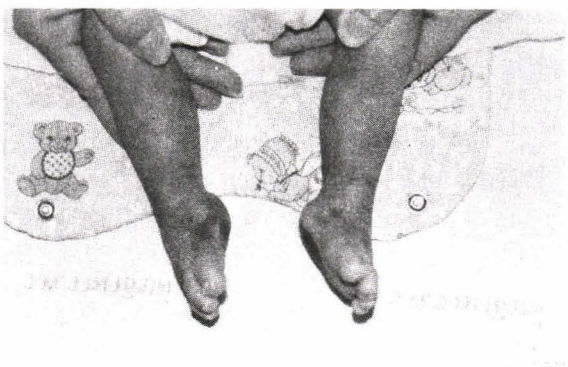

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Fig. 2. Showing the bilateral clubfoot deformities, A. the adduction of forefeet and varus of the mid foot. B. the equinus of both feet with the contracture of tendo achilles.

The initial clinical examination showed the typical findings of Down's syndrome including microbrachycephaly, up-slanted eyes with epicanthal folds, protruding tongue, short neck, lax skin, hypotonia and transverse palmar creases (Fig. 1).

The Trypsin - Giemsa chromosome banding analysis revealed the 46XX, female pattern with translocation at the long arm of chromosome 21. The finding was compatable with the translocation type of Down's syndrome.

From the echocardiographic study, there were mild tricuspid valve regurgitation, mild right pulmonary artery stenosis and persistent foramen ovale.

Clinical examination of her feet revealed the typical appearance of rigid club foot including the adduction of forefeet, fixed equinus of the feet and varus deformities of the mid foot both sides (Fig. 2). The treatment with manipulation and serail casting was started immediately after the initial visit. At the age of 4 months the deformities of the forefoot and mid foot were almost completely corrected. Lengthening of the tendo achilles was performed at the age of 6 months to correct the fixed equinus deformities of both feet. The results were satisfactory as both feet were in neutral position. Nevertheless, the hypotonia was so severe that she could have only sitting balance at the age of 18 months.

DISCUSSION

One of the common foot problems in Down's syndrome is the severe flexible pes planus which is the result of the ligamentous hyperlaxity (1,5). On the contrary, the basic pathology of clubfoot is the contracture of the tendon and ligament surrounding the talocalcaneonavicular ball and socket joint complex. The combination of Down's syndrome and clubfoot appears improbable. In reviewing the literature we found only two references in which these two conditions were presented together(4,6). Although Livingstone and Hirst were the first authors to report two cases of Down's syndrome with equinovarus feet(4), there were no clinical details in these cases. Miller et al reported 8 cases of Down's syndrome who presented with clubfoot deformities(6). Six out of eight cases had completed chromosome study. All of these eight cases eventually required surgical treatment to correct the deformities.

We support the study of Miller et al particularly the treatment as in our patient who eventually required surgical correction too[6].

With regard to the chromosome study, five cases of Miller et al had trisomy 21 and the other one had mosaic pattern. No case with translocation type was found. In contrast, the translocation at the long arm of chromosome 21 was determined in our case.

This presented case confirms that this improbable condition of Down's syndrome and clubfoot does exist. Our patient will be the ninth case. We have a different result of chromosome study from the previous report. Therefore, clubfoot deformity can occur in any type of chromosome abnormality of Down's syndrome.

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กลุ่มอาการดาวน์ ซึ่งมาด้วยอาการผิดรูปร่างแบบเท้าปุก : รายงานผู้ป่วย 1 ราย

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รายงานผู้ป่วยเด็ก 1 ราย มาพบแพทย์ด้วยปัญหาเท้าปุกทั้งสองข้างตั้งแต่แรกเกิด การตรวจร่างกายทั่วไปและการตรวจทางพันธุกรรม ยืนยันการวินิจฉัยว่าเป็นกลุ่มอาการ Down's ซึ่งกลุ่มอาการนี้พบร่วมกับเท้าปุกน้อยมาก เนื่องจากพื้นฐานทางพยาธิวิทยาของทั้งสองโรคต่างกันโดยสิ้นเชิง ในผู้ป่วยรายนี้พบว่าผลการตรวจทางพันธุกรรมเป็นแบบ translocation ซึ่งยังไม่เคยมีรายงานมาก่อน

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