



Recalcitrant scoliosis in Proteus syndrome

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Proteus syndrome is a rare congenital condition with various clinical features such as hemihypertrophy, macrodactyly, subcutaneous masses, brain-like hyperplasia of the soles and/or palms, epidermal naevi and scoliosis with other mesodermal malformations. Multifocal overgrowth can affect various tissues causing severe functional and cosmetic disability, but intellectual and language development are mostly normal. Orthopaedic problems include macrodactyly, hindfoot deformity, limb length inequality, genu valgum and scoliosis. Usually, scoliosis does not respond to bracing, and surgical intervention may be required. Despite surgical correction, instrumentation and fusion, progression of deformity can occur. The authors describe a case with a Th7-L2 scoliosis, which completely relapsed, 20 months after posterior instrumented fusion. Surgery should not be undertaken lightly, given the abnormal growth potency typical for Proteus syndrome.

INTRODUCTION

Proteus syndrome is a rare congenital hamartomatous malformation with a wide spectrum of abnormalities, including overgrowth of various tissues. It was first recognized by Cohen and Hayden in 1979 (4). Biesecker *et al* (1) set up diagnostic criteria: general, mandatory criteria were mosaic distribution of the lesions, progressive course and sporadic occurrence; specific criteria, or category signs, were also necessary: either one sign from category A, or two from category B, or three from category C.

Wiedemann *et al* (12) were the first to coin the term "Proteus syndrome", because of its divergent features, referring to the Greek sea-god who had the ability to appear in any form, indicating the polymorphism of the entity. More than 100 cases with their clinical and radiological features have been reported in the literature (3, 8). Although spinal deformities were present in more than half of the cases, surgical correction of these deformities, especially scoliosis surgery, has rarely been documented (5). The purpose of this study was to report a new case of Proteus syndrome with scoliosis.

CASE REPORT

A 12-year-old girl was seen with "back asymmetry" as her chief complaint, in January 2003. She appeared normal when she was born after a 32-week gestation period. She was the second child of healthy non-consanguineous parents. There was no family history of similar abnormalities. There was no antenatal exposure to medication or radiation.

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Fig. 1. — Thoracolumbar curve Th7-L2, 44°

The intranatal and perinatal periods were uneventful. Birth weight was 2.5 kg. Her family noted overgrowth of her right fourth and fifth toes at 4 months of age. By 8 years of age a lipoma was removed from her back. At the same time her family noticed a spinal curvature, which gradually increased.

On physical examination a left rib hump, shoulder asymmetry and pelvic tilt were noted. Her height was 156 cm, her weight 46 kg, her head circumference 54 cm, all of which were in the normal range for the age. The motor, intellectual and language development were normal. The right fourth and fifth toes were markedly hypertrophic. There was hemihypertrophy of the left leg, which was 6 cm longer than the contralateral leg. A naevus



Fig. 2. — Complete relapse, 20 months after posterior fusion with Cotrel-Dubousset instrumentation.

and a subcutaneous mass, respectively on the left shoulder and in the upper thoracic region, were seen. A sacral haemangioma was also noted. Radiographic examination revealed a 44° left convex thoracolumbar scoliosis Th7-L2, King-Moe type III (fig 1). Ultrasonographic examination of the abdomen revealed bilateral nephrolithiasis. Serum calcium, phosphorus, and alkaline phosphatase were within normal limits.

The diagnosis of Proteus syndrome was made, as the following criteria of Biesecker *et al* (1) were met: mosaic distribution, progressive course and sporadic occurrence (general mandatory criteria); connective tissue naevi on the plantar surface of the left foot (one specific criterion category A is sufficient), overgrowth (specific criterion category B) and presence of lipoma (specific criterion category C).

As bracing was ineffective, a posterior Cotrel-Dubousset fusion Th6-L3 was performed, which reduced the curve to 22°. Unfortunately, at 20 months follow-up the curve had relapsed completely (fig 2). This was probably due to the overgrowth potential of the tissues.

DISCUSSION

Proteus syndrome appears to be due to dysplasia of ectodermal and mesodermal layers, but the exact cause is unknown (2). All cases appear to be sporadic without evidence of an inheritance pattern (10) or sexual bias. Somatic alteration of a dominant lethal gene has been suggested to play a role, resulting in disturbed regulation of tissue growth (7). Regulators of tissue growth may be altered such as growth factor receptors, causing overgrowth of cellular elements of various mesodermal tissues (11). This tendency for overgrowth usually ceases at puberty. The patients should be followed up regularly because of the progressive course of the condition. Since the intelligence and life span are expected to be normal, corrective surgery should be performed when required, but with caution, as recurrence is possible. At birth the child may have no characteristic features of Proteus syndrome. As in our patient, skeletal malformations may develop with years (11). Incomplete forms of the syndrome can also be expected. Growth asymmetry is the most common feature of Proteus syndrome, in varying severity, most commonly involving hands and feet.

The diagnosis of Proteus syndrome is entirely clinical and quite difficult because of the great variability of clinical presentations. The criteria of Biesecker *et al* (1) were used to establish the diagnosis in the case described above. The criteria of Wiedemann *et al* (12) include partial gigantism of hands and/or feet, pigmented naevi, hemihypertrophy, subcutaneous tumours, skull anomalies, accelerated growth and visceral abnormalities. The five major criteria of Samlaska *et al* (10) are : hemihypertrophy, macrodactyly, subcutaneous masses, plantar or palmar masses, exostoses, epidermal naevi and scoliosis.

Systemic involvement is rare, and includes cystic malformations of the lungs (12), brain malformations (9) and ambiguous genitalia (6). Nephrolithiasis was found in our patient, but was not to be seen as essential. The differential diagnosis includes, among others, neurofibromatosis, Klippel-Trenaunay-Weber syndrome, enchondromatosis, Mafucci's syndrome, and Bannayan syndrome (10).

Demetriades *et al* (5) reported a comparable case of Proteus syndrome with progressive scoliosis, also treated surgically. Postoperatively, kyphosis occurred cephalad to the fused area. We conclude that the spinal deformities seen in Proteus syndrome have a high risk of progression, even after adequate surgery. The overgrowth potential of the tissues may be responsible. Although affected individuals have a normal intelligence and life span, surgical correction of spinal deformities in Proteus syndrome should only be considered in the presence of lung or other vital organ dysfunction.

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