



Correction of polydactyly in patients with Ellis-van Creveld syndrome : A report of two cases

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Ellis – van Creveld syndrome (EVC) is a rare chondroectodermal dysplasia presenting several skeletal manifestations and congenital heart malformations. Polydactyly is the most frequent skeletal anomaly. The authors report two cases of EVC syndrome with different manifestations, which underwent surgical treatment for polydactyly.

Keywords : Ellis-van Creveld ; polydactyly ; congenital anomalies.

Ellis-van Creveld syndrome (EVC) is a rare autosomal recessive congenital disease. It was first described in 1940 by Ellis and van Creveld (2). The syndrome is characterised by chondroectodermal dysplasia, polydactyly and congenital heart malformations. Ectodermal dysplasia is characterised by friable nails, abnormal teeth and alopecia. The chondrodysplasia is characterized by disproportionate small stature, and a variety of skeletal anomalies of which polydactyly is the most frequent. This paper reports two cases of EVC syndrome, with focus on the orthopaedic management.

CASE 1

An Algerian boy from a consanguineous marriage was born with respiratory distress and bilateral postaxial polydactyly of the hands and feet. At 2 years of age, weight, height and skull perimeter were below P3. The infant was not

walking yet. He presented a long and narrow thorax with short stature due to shortness of the lower limbs.

Extra-orthopaedic features

The heart malformation was an atrial septal defect extending to the mitral valve. The heart defect was responsible for growth disturbance and multiple respiratory infections. Radiographs showed a large heart in a narrow thoracic cage ; electrocardiogram (ECG) showed right heart hypertrophy. Cardiac surgery was done by closure of the septal defect and remodelling of the mitral valve.

Orthopaedic features

A harmonious hexadactyly of the four extremities was the major problem. The postaxial fingers were not functional and the parents requested a surgical resection of the extra fingers. The radiographs showed a complete sixth metacarpal and metatarsal

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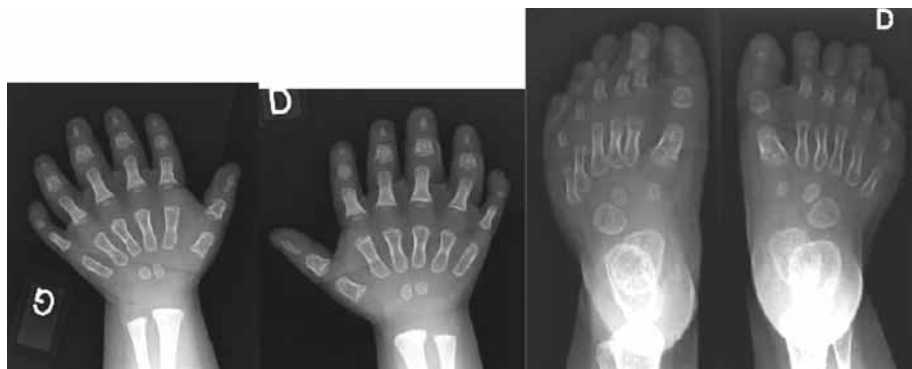


Fig. 1. — Radiographs showing a complete 6th ray with hypoplastic phalanges in both hands and feet.

bone including the first phalange. At the hands, a partial second phalange was also present. The 6th metacarpals were shorter; phalanges were hypoplastic, especially in the upper limbs (fig 1).

Two surgical teams (a hand surgeon and a paediatric surgeon) worked together to correct the hands and feet polydactyly in the same session. The hand surgeon removed the lateral finger and the 6th metacarpal bone on both sides and transferred the ulnar collateral ligament and abductor digiti quinti to the fifth ray. The paediatric surgeon disarticulated the lateral ray and toe from the cuboid bones using a zigzag incision to minimise fibrotic retraction.

CASE 2

A girl, born from a non-consanguineous union, after a normal pregnancy, presented with an EVC syndrome. At birth, the child had shortening of the limbs, bilateral polydactyly with hypoplastic nails, a narrow thorax, oral anomalies and a congenital heart malformation.

Extra-orthopaedic features

A hypoplastic left heart syndrome with a major atrial septal defect explained the dyspnea and cyanosis, especially when the child cried. The defect of the mitral valve was minor. At two years of age, the heart malformation was operated using a pericardial patch to separate both atria, following which the girl had a better physical condition despite a small mitral regurgitation.

The oral manifestations included notching of the alveolar ridge, labio-gingival adhesions and hypodontia involving the maxillary region. These anomalies are important risk factors for oral infections.

Orthopaedic features

In this case, the polydactyly was complex. The 2nd and 6th fingers of the right hand were hypoplastic. The left hand showed a synpolydactyly of the 5th and 6th fingers and a rudimentary 7th finger. The nails were hypoplastic. Radiographs showed hypoplastic phalanges in the lateral rays, carpal fusion, and a complex syndactyly of the left hand (fig 2). There were no associated foot malformations.

Hand surgery was performed when the girl was between 3 and 4 years of age. On the right hand, the 2nd finger was more hypoplastic than the 6th. It was decided to perform resection of the 2nd ray with transposition of the adjacent intermetacarpal liga-



Fig. 2. — Radiographs showing carpal fusion, complex syndactyly and polydactyly.

ment, covered by a skin flap. On the left side, the incomplete syndactyly of the 1st web space was corrected by a Z-plasty and a skin graft from the ulnar side of the hand. Resection of the 6th and 7th rays was then performed, taking care to separate the metacarpal and the phalange from the 5th ray (fig 3).



Fig. 3. — Complex polydactyly of both hands : pre- and post-operative view..

DISCUSSION

Chondroectodermal dysplasia, or Ellis-van Creveld syndrome, is a rare autosomal recessive congenital anomaly. The incidence is around 1/60.000 births, but frequency increases in case of consanguineous union. The mutation is located on chromosome 4, but two different genes have been found responsible so far (5,6). Clinical diagnosis is made at birth, but can be made prenatally by a morphologic ultrasound examination, showing intrauterine growth retardation and cardiac defect (1, 8).

There are several skeletal manifestations. A long narrow chest is frequently seen, with short poorly-developed ribs giving rise to a pigeon breast and respiratory difficulties. Acromelic and mesomelic shortness of limbs is often encountered. At the upper limb, a curvature of the proximal humerus may be present, the radial head may be dislocated, and the ulna is hypoplastic. At the wrist and hand, a variety of skeletal anomalies may be observed, including fusion of the hamate and capitate bones, clinodactyly of the 5th finger, fusion of the 5th and 6th metacarpals, and disturbance in bone modelling of the metacarpals and phalanges (4). At the lower limb, pelvic dysplasia is frequent, with a horizontal

acetabular roof with lateral spikes. A defect of the lateral aspect of the proximal tibia produces a genu valgum deformity and patella dislocation. The fibula is sometimes shorter than the tibia, resulting in a calcaneo-valgus deformity.

Congenital heart malformations occur in 50% of cases ; the most frequent manifestation is the single atrium (7). The presence of congenital heart disease imposes surgical correction and appears to be the main determinant of life expectancy. Dental and oral manifestations often occur in Ellis-van Creveld syndrome. Eye, genital and renal anomalies may also occur, but are very rare (9).

In conclusion, a multidisciplinary approach is advocated for a complete management of a patient with chondroectodermal dysplasia. The surgical technique for correction of polydactyly should be tailored to the individual case.

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