ORIGINAL ARTICLE

Control cases of extreme cleft hand and feet deformities in three generations

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***SUMMARY*:** We present a family of four affected individuals in three generations with extreme clefting of the hands and feet anomalies. The lack of interest for medical treatment of the individuals is appraised here in the context of the current strategies of management.

Key Words: cleft hand, congenital anomaly

The International Federation of Surgical Societies of the Hand (IFSSH) in one series gave a minimum estimate of 22.91 limb deficiencies occurring per 10,000 births with radial club hand representing 1.3.[1] In another population based Hungarian study 17% cases of limb deficiencies were related to genetic disorders.[2] The currently used classification is based on that of Swanson modified by IFSSH in 1983,[3,4,5] though from time to time non-classifiable cases are still reported from every part of the globe.[6,7,8]

Limb buds may be considered a single developmental field with the rostral set (arms) developing somewhat earlier than the caudal (legs).[9] Digit formation is apparently controlled by the zone of polarizing activity which releases a diffusible morphogen specifying positional information along the anterior/posterior axis.[10] Vertebrates have 39 HOX genes organized into four clusters with major roles in development. A variety of limb malformations in human beings are now known to be caused by chromosomal deletions involving HOXD and HOXA clusters, specific and regulatory mutations in HOXD 13 and HOXA 13 genes.[11] The environmental factors, teratogens induced,[12] and genetic factors both can cause limb defects.

The following case report illustrates the aspects mentioned above.

Case Report

A fifty-year-old lady with a thirty year old daughter and eight and six year old grandsons were seen in our clinic incidentally while they were visiting another patient. All of them showed an identical deformity in all limbs. The hands comprised of a single digit present on the ulnar side. The digit was rotated radially and the rest of the hand, though smaller in breadth, was present up to the palm. The feet also comprised of only one digit present on the fibular side. The sensory and motor functions in all the limbs were normal.

They were all products of uncomplicated full term pregnancy, labour and normal vaginal delivery. They were found to have normal height and weight and systemic examination was normal.

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Radiological examination revealed an ulnar based finger with two metacarpals, absent trapezium but normal wrist and distal radio ulnar joints and radius and ulna in both hands. In their feet a single fibular digit and a fibular metatarsal were present with absent cuneiform bones.

A diagnosis of failure of formation presenting as a variant of bilateral longitudinal radial ray deficiency in hands and bilateral tibial ray deficiency in feet was made. The identical deformity in three generations indicated the expression of an autosomal dominant gene.

Functionally, the ability to hold both small and large objects with their hands unilaterally and with both hands combined was very good and the gait was normal. The family allowed the examination but refused the option of surgical treatment for any member of the family.

Discussion

The above family presents with uniform expression of extreme clefting in all limbs, with a solitary ulnar digit in the hands and a solitary fibular digit in the feet in all three generations, suggesting an autosomal dominant mode of inheritance. Surgeons have improved their armamentarium with microvascular tissue transfer and bone lengthening techniques.[13] Increasing experience with timing and selection of surgical techniques has helped in decision-making and improved outcome.[14]

Together with counselling, occupational therapy and social support services, it is now possible to achieve what was once unimaginable.

Antenatal ultrasound, analgesics, amniotic fluid and chorionic villus sampling have allowed malformations to be predicted[15], allowing parents to be mentally prepared of the outcome of the pregnancy, or offer them the option of termination. Genetic counselling can be offered at this stage as a useful guide for future pregnancies.

Such detailed management plans for these patients emphasize how western society sees such malformations as having profound implications on the physical, psychological, and social welfare of the individual.

On the other hand, the refusal to seek medical help by the reported individuals verifies the fact that adaptation to the physical limb deformities does occur within a cohesive family based unit. It is salutatory that the acceptance of the deformities within the family has produced relatively normal functioning individuals and calls into question the need for specialised surgical intervention in current western society.

The above-mentioned case puts into question the plans to follow the long and arduous path of surgical intervention to correct these deformities, or even considering termination of a pregnancy. Control groups of patients who have had absolutely no surgical intervention can play an important role here, with whom the outcomes of surgery can be compared, to see whether a tangible difference to the lives of these people is really made or not? Are these patients offered an improvement in quality of life, the extent of which is largely unknown due to the lack of control cases, at the expense of a protracted surgical and rehabilitation process and improved cosmesis?

It may also be argued that in western society the social stigma attached to such deformities would make the option of no surgical intervention unacceptable, and that only in certain sub-cultures may this practice of non-intervention be acceptable, perhaps even recommended?

Perhaps, a better way to deal with these deformities is to reassure and support the parents and the child, with the idea that a deformed but functioning hand is better than a scarred, stiff, and possibly less satisfactorily functioning hand, which will still look deformed[16].

Fig 1

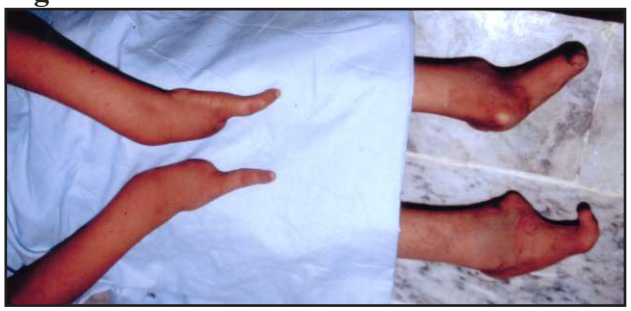

Fig 2a

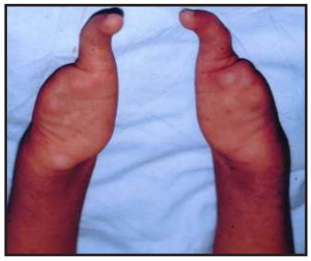

Fig 2b

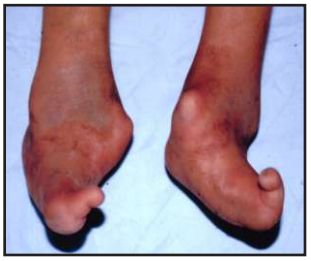

Fig 2c

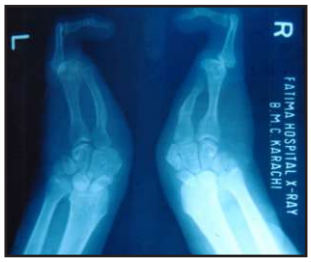

Fig 2d

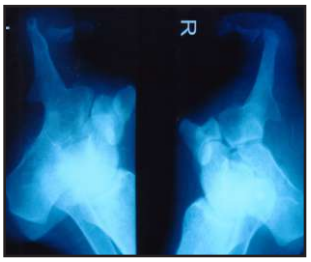

Fig 2e

Fig 3a

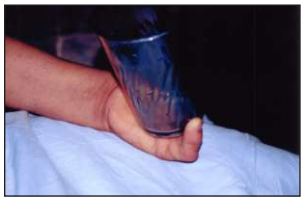

Fig 3b

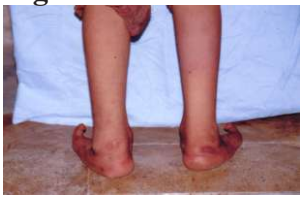

Fig 3c

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