True Symphalangism (Absence of Proximal Interphalangeal Joints)

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ABSTRACT

Cases recognized and confirmed as of Proximal Symphalangism due to human noggin (NOG) mutation. With the use of chromosomal study (Routine Cytogenetic test) (karyotyping) and radiologic examination, along with collection of detailed family history information make it possible to describe the morphology and anatomical variation to the previously reported anomalies of congenital nature.

Keywords: Symphalangism, Absence of Proximal Interphalangeal (PIP) Joint, Distal Interphalangeal (DIP) Joint, Human noggin (NOG)

1. Introduction

Congenital anomalies of the fingers are uncommon and a number of different types have been described in the literatures. Some, such as webbing of the fingers, are mentioned in standard medical textbooks. However, Symphalangism is a rare one and it was therefore, thought worthwhile to report its morphology and the family history in some detail. In spite of that such anomaly might lead to some functional disability, yet it does not need surgical treatment in all cases. This condition was found with two siblings without evidence of any similar anomalies in their parents (fig. 1), patients did not complain of any functional disability in their daily activities.
Fig. 1, pedigree of cases of absence of proximal Interphalangeal joints.

2. Methodology

Case no.1
On 18th May 1972 this anomaly was found by chance in a 20 year old male, who sought advice on another unrelated matter. On examination of his hands, it was found that the ulnar four fingers were narrower than normal in the region, where the proximal two phalanges would normally be found and there were no skin creases corresponding to the proximal Interphalangeal joints on the dorsal and palmar sides, the skin was perfectly smooth. Creases were present in the region of the distal Interphalangeal joints and the terminal segment of the fingers, including the nails appeared normal (fig.2, 3 &4). Flexion was possible at the DIP joints. In case of the thumbs the interphalangeal creases over palmar and dorsal aspects were absent. An exactly similar anomaly was found in the lateral four toes, although it was less noticeable. On clinical examination, no other abnormalities could be found.
RADIOLOGICAL EXAMINATION
A radiological examination of the hands showed that the distal phalanx appeared to be normal, as did the distal Interphalangeal joint. The proximal two phalanges of the four ulnar fingers were replaced by a single apparently normal phalanx, which its length was approximately the length of the two missing phalanges. There was no sign of the proximal Interphalangeal joints. In the case of the thumbs, there were partial fusion between the two phalanges, but otherwise the bones appeared normal. (Fig. 5 & 6).

Radiology of the feet confirmed the absence of the proximal Interphalangeal joints at the lateral four toes and the big toes showed a similar anomaly to the thumbs.
LABORATORY TESTS

A routine laboratory test that has been done includes Full Blood Count, Renal Function Tests, and Serum calcium were all within normal limits. Also a chromosomal study Routine Cytogenetic test (karyotyping) by a modification procedure described by Moorhead ET al had been performed and showed no further abnormalities (fig.12).

![Image of karyotype](image1)

Fig.12. Case No.1, Metaphase and Karyotype

CASE NO.2

This patient was the brother of Case no.1. He was 29 years old, married with normal children. On examination both hands and feet showed abnormalities of the four ulnar digits exactly similar to Case no.1. In case of thumbs, a slight creasing on the palmar and dorsal surfaces opposite the position of normal interphalangeal joints was found. (fig.7, 8 & 9)

![Image of hands](image2)

![Image of hands](image3)

Fig.7. Case No.2, Dorsal side of hands  
Fig.8. Case No.2, Palmar side of hands
**Radiological Examination**

The findings were similar to case no.1, the Interphalangeal joints of the thumbs showed complete joint spaces between the two phalanges. In the feet there were only two phalanges in the second and third toes, with absence of the proximal Interphalangeal joints, but in the fourth and fifth toes there were a very narrow joint space present in the region of the proximal Interphalangeal joints, but on examination of these toes there was limitation of movement. Routine examinations showed no abnormalities elsewhere in the body (fig.10 & 11).

**Laboratory Tests**

A routine laboratory test that has been done includes Full Blood Count, Renal Function Tests, and Serum calcium were all within normal limits. Also a chromosomal study **Routine Cytogenetic test** (karyotyping) by a modification procedure described by Moorhead et al. (1960) has been performed and showed no further abnormalities (fig.13)
Family History
The pedigree of the above cases shows, that the maternal and paternal grandfathers, who were first cousins, had both suffered from the same condition (fig.1) the father and mother, however, were normal. An informed consent was signed by both patients for the use of photographic evidence for medical purposes.

3. Results & Discussion

Symphalangism is an autosomal dominant disorder, the pedigree pattern of our cases also reflects this assumption (fig.1). It was first described by Harvey Cushing in the first published issue of Genetics (Cushing, 1919).
The proximal Symphalangism are more common than the distal type (Diganta et al., 2006). It seems that in most of the reviewed literatures the affected persons were males, like in our cases. In the reviewed literatures it was said that the PIP or the DIP joints were replaced by bony or fibrous ankylosed joints (Cushing, 1919; Diganta et al., 2006; Charles et al., 1994; Krohn et al., 1989) while in our case No. 1, the proximal two phalanges in the four ulnar fingers were replaced by a single apparently normal phalanx, whose length was approximately the length of the two missing phalanges. There was no sign of the proximal Interphalangeal joint. In the case of thumbs, there were partial fusion of the two bones, but otherwise the bones appeared normal (Fig. 5 & 6).

While in the second case the findings were similar to the first case, except that in case of thumbs the interphalangeal skin creases were present and the X-Ray of the hands showed no fusion. There were no signs of osteoarthritis or even marked disability in our cases at the time of presentation, as it has been mentioned that osteoarthritis in the proximal and distal joints to the fused joints has been reported (Krohn et al., 1989). Now with the advance in molecular research it became possible to define these cases of Proximal Symphalangism due to Human noggin (NOG) mutations (Athanasakis et al., 2012; Usami et al., 2012).

4. Conclusion

Current development in the molecular research make possible to recognize cases of Proximal Symphalangism due to human noggin (NOG) mutation, although it was not possible in the past to define and recognize the actual genuine cause of these kinds of congenital anomalies.

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References


