Mesoaxial synostotic syndactyly with phalangeal reduction (MSSD): syndactyly type IX

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Letter to editor:

We read with great interest the case presented by Dr. Weinrich et al. [1] about novel nonsyndromic unilateral syndactyly of the hand. We would like to point out that the condition with fusion of mesoaxial digital rays culminating in four fingers has been characterized as ‘syndactyly type IX’ (OMIM 609432) [2]. This entity, also known as mesoaxial synostotic syndactyly with phalangeal reduction (MSSD; Malik–Percin type), is a very rare limb anomaly with only six cases reported to date [3]. The malformation is characterized by bony fusion at the metacarpal level, resulting in a four-fingered hand. Usually, the 3rd to 4th metacarpals are involved in the fusion but there can be a fusion between the 2nd and 3rd metacarpals. There can be concomitant fusion between the 3rd and 4th metacarpals in one hand and between the 2nd and 3rd metacarpals in other hand in the same individual. The fused fingers appear as a single hypertrophic digit with symphalangism of terminal phalanges. Further, there is mild hypoplasia of distal phalanges, which is more obvious in the preaxial and postaxial digits. The thumbs are usually reduced in size and there is clinodactyly of the 5th fingers with symphalangism. The feet are only mildly affected and are presented with cutaneous fusion of preaxial or mesoaxial toes and terminal phalangeal hypoplasia of all toes [4, 5]. The condition is non-progressive and is not associated with any pain. There is, however, a slight reduction in hand size and its strength and grasp may be compromised. Further, symphalangism of the accompanying fingers may cause difficulty in bending the joints. Bilateral presentation is common but unilateral cases are also known

(unpublished data). This anomaly has been presented as an isolated limb malformation. It segregates in an autosomal recessive manner and five of the six reported families originate from cultures where consanguinity is highly conspicuous. We have previously shown that type IX syndactyly is caused by mutation in *BHLHA9* gene [3].

Therefore, based on our clinical experience, we conclude that the case presented by Weinrich et al. is not novel, and may be classified as syndactyly type IX (MSSD).

Yours sincerely,

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