

CASE REPORT

Fanconi Anemia Concurrent with an Unusual Thumb Polydactyly: A Case Report

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*Research performed at Urmia University of Medical Sciences, Urmia, Iran**Received: 8 April 2015**Accepted: 17 August 2015***Abstract**

This case report presents a case of Fanconi's Anemia with an unusual thumb polydactyly in a 2-year-old boy. The extra thumb had no nail, nail bed and distal phalanx. The extra thumb had no active motion. The duplication of the thumb occurred at the carpometacarpal joint but its morphology did not match with any classification described for thumb polydactyly. Although his thumb polydactyly was apparent at birth, Fanconi's anemia was not suspected until during a routine pre-operative laboratory test (CBC) for the elective surgery of his thumb. An early diagnosis of FA is important and the hand surgeons may be the first to have the opportunity to suspect and diagnose the underlying life threatening disorder. This case report presented an opportunity to diagnose a fatal disorder by a routine pre operative laboratory test. To the best of my knowledge, the phenotype of the thumb polydactyly of the current case has not yet reported.

Key Words: Congenital anomaly, Fanconi anemia, Thumb duplication, Thumb polydactyly

Introduction

Fanconi's Anemia (FA) is an inherited disorder with an autosomal recessive inheritance. FA has a rare incidence but it has serious hematologic consequences. Because of a defect in DNA repair, FA predisposes the patients to progressive bone marrow failure and development of aplastic anemia, leukemia and solid tumors (1- 3).

Thumb polydactyly is a common hand anomaly but its concurrence with FA is a very rare (4, 5). This case report presents a case of FA concurrent with an unusual thumb polydactyly which morphologically did not match with any classifications described for the thumb polydactyly.

Case Report

A 2-year-old boy was referred because of his right thumb polydactyly [Figure 1]. His parents were first cousins. On clinical examination the most radial thumb was underdeveloped. The extra radial thumb had an abducted position probably because of the unbalanced pull of the abductor pollicis longus attached to the base of the most radial thumb. The

extra thumb had no nail, nail bed and distal phalanx. The extra thumb had no active motion. The thenar eminence was flat [Figure 2] and there was no other obvious visceral and skeletal abnormality.

On plain radiograph, the thumb polydactyly was at the level of carpometacarpal joint. The most radial thumb had only two osseous components; the proximal bone represented the thumb metacarpal bone and the distal bone represented the proximal phalanx [Figure 3]. The distal phalanx was absent. The forearm bones were normal.

Upon preoperative routine blood testing for an elective surgery for ablation of the extra thumb, blood cell count demonstrated anemia and thrombocytopenia with a platelet count of 55000 (reference range: 100000-400000). The blood cell count abnormality raised the suspicion of an underlying hematologic disorder. Peripheral blood smear demonstrated hypocellularity. Cytogenetic breakage study demonstrated that the mean chromosomal breakage was considerably increased which was a cytogenetic evidence of FA.

The patient was scheduled for hematopoietic stem cell transplant; his parents postponed his hand

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Figure 1. Dorsal view of the right hand with thumb polydactyly. The nail is absent.

surgery until he had recovered from his stem cell transplant.

Discussion

FA has variable phenotypes. FA may be associated with growth retardation, short stature, skin hyperpigmentation, skeletal abnormalities, central nervous system defects, urogenital malformations, eye, ear and maxillofacial defects, gastrointestinal defects, cardiopulmonary abnormalities as well as the progressive pancytopenia; 25% of FA patients, however may have no other congenital abnormalities (1-3, 6, 7).

The age of onset of hematologic abnormalities may be seen variable with the mean age reported around 8 years. Initially the blood cell count may be adequate for an elective surgery; however, a downward trend



Figure 3. Anteroposterior radiograph of the right hand with thumb polydactyly demonstrates that thumb polydactyly was at the level of carpometacarpal joint. In the most radial thumb the proximal bone represents the thumb metacarpal bone and the distal bone represents the proximal phalanx.



Figure 2. Palmar view of the right hand with thumb polydactyly. The thenar eminence is flat.

may also be noted. Hematopoietic stem cell transplant should ideally proceed before the need for regular transfusions. Therefore, an early diagnosis of FA is important and the hand surgeons may be the first to have the opportunity to suspect and diagnose the underlying life threatening disorder (1, 2, 5, 7).

In a cumulative data of 700 patients with FA reported by Alter, 50% of the patients had preaxial dysplasias; 300 had thumb anomalies including absent, hypoplastic, floating, bifid, supernumerary, triphalangeal thumbs and hypoplastic thenar eminence. The association of radial longitudinal deficiency and thumb hypoplasia with FA has been well documented. Wilks and Alter have suggested that there may be an association between thumb duplication and FA (2, 5).

Thumb polydactyly classifications are based on the radiographs of the osseous structures. The complexity of thumb polydactyly in the current case is not fully described by the Wassel classification or the elaborated classification by Zuidam (8, 9). The ossification center of the trapezium does not appear until 5 years old. In the current case, it is not possible to comment on the extent of the duplication to the trapezium or other carpal abnormality.

This case report reminds us that a fatal disorder may be diagnosed by a routine pre-operative laboratory test. In the current case report Fanconi anemia has been seen with a thumb polydactyly. However, to the best of my knowledge, the phenotype of the thumb polydactyly of the current case has not yet reported. Hopefully, piling up the case reports may provide clues about association of Fanconi anemia and thumb polydactyly in future.

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