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Fanconi Anemia with Triphalangeal Thumbs, Syndactyly and Contractures of the Fingers in a 2 Year Old Boy

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KEY WORDS Fanconi anemia; syndactyly; contractures; triphalangeal thumbs; chromosomal breakage.

ABSTRACT Fanconi Anemia (FA) is a rare autosomal recessive disorder associated with pancytopenia, spontaneous chromosome instability and a variety of congenital anomalies. Hypersensitivity to bifunctional alkylating or DNA crosslinking agents like Mitomycin C (MMC), Diepoxybutane (DEB) and Nitrogen Mustard (HN_2) is used as a differential diagnostic test. A variable phenotype and age of onset of anemia make diagnosis difficult in some cases. We report a case of Fanconi anemia detected by the MMC stress test in a 2 year old boy, operated for bilateral syndactyly and contractures of fingers. He had a bifid thumb on the left hand and bilateral triphalangeal thumbs. There was no history of consanguinity or malformations, though a maternal uncle had a bifid thumb. USG in a subsequent pregnancy showed bony anomalies like scoliosis, talipes, contractures and radial aplasia, consistent with FA. The parents opted for termination. An early diagnosis of FA in a non-manifesting child would provide more time to explore different treatment options, since a delay in diagnosis could have serious consequences.

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