Original Paper

Chromosomal aberrations in patients suspected with the risk of Fanconi anemia

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Abstract

Background and Objective: Fanconi anemia is the most prevalent inherited aplastic anemia. Diagnosis based solely on the recognition of clinical symptoms is not reliable. This study was done to determine chromosomal aberrations in patients suspected with the risk of Fanconi anemia in the Eastern Azarbaijan province- Iran.

Materials and Methods: This descriptive study was conducted on 20 patients in the Eastern Azarbaijan province-Iran. The cytogenetic method was used to determine type and number of chromosomal disorders.

Results: Nine eight and nine patients had co-morbid anemia, platelet deficiency and 9 patients had hand and finger deformities, respectively. Using cytogenetic method, Fanconi anemia was confirmed in 5 (25%) of the cases. The percentage of mitotic abnormalities in the chromosomes without administration of mitomycin C varied between 5-30% in the cultures of the 5 affected and between 0-4% in the 15 unaffected patients with the administration of mitomycin C, the percentages were increased up to 35-78% and 0-20% in affected and unaffected patients, respectively.

Conclusion: Fanconi anemia is confirmed precisely in 25% of suspected patients using cytogenetic method.

Keywords: Fanconi anemia, Chromosomal breakage, Mitomycin C

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