J Blood Disord Transfus 2018,Volume:9
DOI: 10.4172/2155-9864-C3-038

Phenotypic presentation of Pakistani Fanconi anemia patients: A single center study

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Fanconi anemia (FA) is the most common inherited bone marrow failure disorder. It is characterized by bone marrow failure, congenital abnormalities and increased propensity to develop malignancies. FA is a chromosomal instability disorder for being error in DNA repair mechanism. As per international Fanconi anemia data base, more than 21 genes or complementary groups have been identified. Clinical presentation of Fanconi anemia is very heterogeneous. In Pakistan, incidence and presentation of Fanconi anemia has never been documented. Total of 60 diagnosed patients of Fanconi anemia (on chromosomal breakage analysis MMC test) were included in the study after taking written and verbal informed consent, of which 33% were female. Majority of (96%) patients presented with pancytopenia (reason most likely that our center is a hematology center), 26% patients had skeletal abnormalities while 23% had renal structural abnormalities. Cafe au lait, microphthalmia and facial abnormalities were present in 41%, 57% and 61% patients respectively. Ninety eight percent (98%) were below 5th centile for both height and weight. Family history was present in 41% patients. The phenotypic presentation of Fanconi anemia is highly variable. Any patient presenting with cytopenia, skeletal defects, abnormal skin pigmentation or atypical faces should be worked up for Fanconi anemia.

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