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FANCG 637-643 DELETION MUTATION: FREQUENCY IN BLACK PATIENTS WITH ACUTE MYELOID LEUKAEMIA OR APLASTIC ANAEMIA AND THE CLINICAL PHENOTYPE OF HOMOZYGOTES

Tabitha Haw

Fanconi anaemia (FA) is an autosomal recessive disorder characterised by aplastic anaemia (AA) and a high risk of developing acute myeloid leukaemia (AML). It is unknown whether heterozygote carriers are also predisposed to developing these disorders.

The black South African population group is ideal for FA mutation screening because the presence of a founder mutation, *FANCG 637-643*, makes screening relatively straight forward. Three individuals with AML (115 screened) and one with AA (78 screened) were found to be heterozygous for the black South African founder mutation. From our data it seems unlikely that this mutation places heterozygous carriers of the mutation at high risk of developing AML or AA. Three children with AA out of 26 screened, were homozygous for the mutation. This finding reiterates the importance of screening all children with AA for FA.

The frequency of certain congenital abnormalities in black South African FA patients was compared to patients described by other research groups. The frequencies of the abnormalities were similar to other FANCG cohorts described but significant differences to a group of FA patients from unspecified complementation groups were found. This difference could be because different complementation groups are associated more or less strongly with specific abnormalities.

WiSETD

It was found previously that particular congenital abnormalities in FA patients are associated with a poor haematological outcome. We concluded that black South African FANCG patients have a high risk of early development of AA even though they do not have a high frequency of congenital abnormalities.

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