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Genetics of Fragile X Syndrome: A Systematic Data from the Indian Population

**G.K. Chetan , K.R. Manjunatha, R. Arathi, P. Latha, S. Padma, G.V. Bhakar
Rao,
H.N. Venkatesh, V. Shobha*, S. Shobha*, S.R. Girimaji* and S. Sheshadri***

Departments of Human Genetics and Psychiatry, National Institute of Mental
Health and Neurosciences, Bangalore 560 029, Karnataka, India*

KEY WORDS Fragile X syndrome; Indian population; frequency; diagnosis; genetic counselling.

ABSTRACT Fragile X syndrome is the commonest form of X-linked disorder. Its frequency among MR ranges from 6-9%. There are a few reports available on Fragile X syndrome from Indian population and we have screened for 300 MR subjects with 26 subjects (8.6%) showing Fragile X chromosome expression in 3-40% of lymphocyte cultures. Herein, we have discussed frequency of Fragile X in the Indian population. Among the subject groups, there were 7 families with multiple sibs being affected and 3 mothers of the affected subjects showed carrier status. The combined data from the Indian population is presented in this study for better understanding of the population dynamics of this syndrome.

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