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Expression of Fragile Xq 27.3 in a Patient with Clinical Features of Deletion 9p Syndrome

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There are reports showing association of fragile X with trisomy 21, Klinefelter syndrome, triple X, Duchenne muscular dystrophy (DMD) and familial psychiatric disorder(l-5). In this report we describe a case of fragile X associated with 'clinical features of deletion 9p syndrome'.

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Case Report

The propositus was a 12-month-old male born to non consanguineous parents at 3rd full term breech delivery. His weight was 7.4 kg, height was 65.5 cm and head circumference was 39 cm. The clinical examinations showed the features of deletion 9p syndrome, *viz.,* trigonocephaly, flat nasal bridge, anteverted nares, long philtrum, wide spaced nipples, micrognathia, short stubby hands and the features of fragile X syndrome like enlarged ears, odd facies *{Fig. la)*. Metacarpal index appeared less (3.8) compared to the normal metacarpal index which is 5.3 for the average age of 12 months *(Fig. lb)*.

Cytogenetics

Cytogenetic analysis with GTG banding to the 800 to 900 band stage revealed 46, XY with both the chromosome 9 to be normal. Studies on fragile X expression was performed using medium TC 199 with 5-fluoro-2'-deoxyuridine (at the concentration of

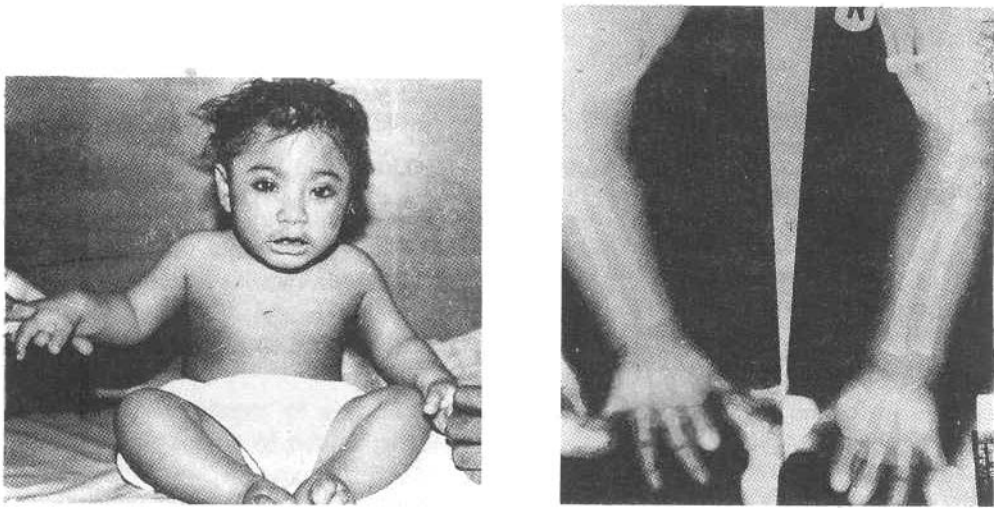

Fig. 1(a). Clinical features in a 12-month-old male patient. (b) Radiological photograph showing metacarpal index of the patient

10-7 M) added 24 h before harvesting. The frequency of expression of fragile Xq27.3 was 4.3% (Fig. 2). Parents and other two siblings were cytogenetically normal. There was no family history of malformation or mental subnormality.

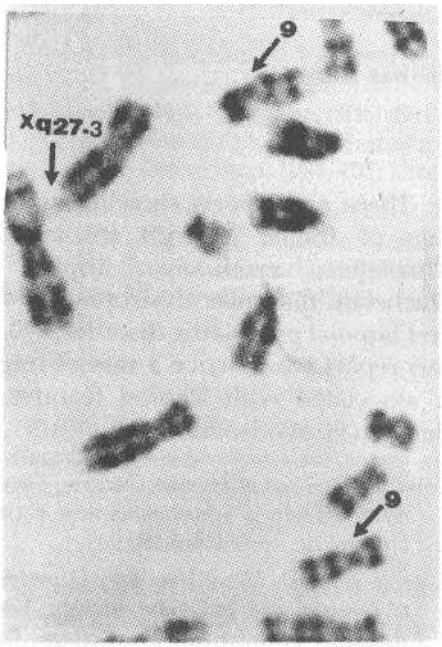

Fig. 2. Partial metaphase showing both norn chromosome 9 and fragile Xq 27.3.

Discussion

To the best of our knowledge this is the first case of fragile X associated with clinical features of deletion 9p syndrome. There are 50,000 genes per haploid set. The 2000 band stage of resolution^) gives an average of 25 genes per band(7). Even at this stage of resolution gene deletion or DNA rearrangement cutting across two genes would be missed cytogenetically. There have been cases of subjects possessing three or four genetic disorders, all known to localize on Xp, in which molecular studies were required to confirm an enormous deletion in DNA terms but a minute,

barely detectable deletion at the cytogenetic level(8,9). We achieved a resolution to the 800 to 900 band stage and at this stage there are an average of 50 genes per band. Tn the present case, the features of deletion 9p syndrome may be due to loss of some few genes resulting in the pafhogenesis which could not be detected cytogenetically. Hence, lack of a detectable difference in banding pattern in any chromosome does not exclude deletion 9p syndrome.

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