HHS Public Access

Author manuscript

Hum Genet. Author manuscript; available in PMC 2017 June 05.

Published in final edited form as:

Hum Genet. 1988 April ; 78(4): 383.

Chromosome breakage in control and fragile X subjects using folate-deficient culture conditions

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The fragile X syndrome is a well-recognized form of X-linked mental retardation with a chromosomal fragile site at Xq27.3 observed in cells grown in folate-deficient culture medium (Brookwell and Turner 1983; Sutherland 1979; Chudley and Hagerman 1987). To determine if generalized chromosome instability exists in fragile X syndrome patients, 5793 lymphocytes from 55 control subjects (33 males and 22 females, average age 29.1 years with a range of 0.1 to 76 years) and 2731 lymphocytes from 27 fragile X syndrome individuals (21 males and 6 females, average age of 27.3 years with a range of 2 to 67 years) were studied in cultures with folate-deficient medium 199. The fragile X chromosome expression ranged from 1% to 55% with an average of 16%. In cells from the control subjects 85 chromosome breaks were observed with a frequency ranging from 0 to 14 (average 1.6) per 100 meta-phases. In cells from the fragile X syndrome patients 41 breaks were seen (excluding the Xq27 site) and the frequency ranged from 0 to 6 (average 1.5) per 100 metaphases. The site of the breaks was determined in banded chromosomes. The 3p14 and 16q23 sites were the most common autosomal locations for breaks in cells from both control and fragile X syndrome individuals. In conclusion our results with a relatively large sample of control and fragile X syndrome individuals confirmed earlier reports (Vekemans et al. 1983; Branda et al. 1984) that no increased chromosome breakage exists in the fragile X syndrome individuals compared with control subjects.

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