A fragile site 10q25 in human sperm chromosomes

During a study of sperm chromosomal abnormalities in normal men,^{1,2} we found a donor who expressed the fragile site at 10q25 in his sperm. Unfortunately, we have not been able to contact him to confirm the presence of the fragile 10q25 in lymphocytes.

The donor was a normal healthy 44 year old male. He and his wife had been infertile for four years although he had fathered one normal child in a previous marriage. A total of 35 Q banded sperm chromosome complements

was obtained using the hamster egg technique previously described.³ Details of the sperm chromosomal analysis are shown in the table. Examples of the fragile 10q25 chromosome in sperm are shown in the figure.

Since four of the 35 sperm complements (11.4%) had a gap or break at 10q25, it is likely that this represents expression of the fragile site in meiotic chromosomes. We have never found a chromosome gap or break repeated in more than one sperm in any subject in over 80 men studied (unpublished observations).

Three of the sperm chromosome complements (8.6%) had a small acentric fragment. These fragments are very rare in human sperm; in 1426 sperm karyotypes from 45 normal men only 0.5% contained such a fragment (unpublished results). This high frequency of chromosomal fragments suggests that the fragile site at 10q25 may predispose to meiotic chromosomal breakage and dissociation of the segment distal to the fragile site.

This report demonstrates that a fragile site is expressed in meiotic chromosomes and that it may be associated with an increased risk of abnormalities. Further studies on other subjects with fragile sites will determine whether these sites do increase the risk of chromosomal breakage and rearrangements.

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TABLE Sperm chromosome complements.

	No of sperm
23,X	13
23,Y	15
23,X.fra(10q25)	1
23,Y.fra(10q25)	2
23,Y.fra(10q25.csg(4q31)	1
23,X.+fragment	1
23,Y.+fragment	2
Total	35

FIGURE Examples of chromosome 10 in sperm without (left) and with (middle and right) the 10q25 fragile site. Top row: solid stain with Giemsa. Bottom row: Q bands with quinacrine dihydrochloride.

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