Two chromosome fragments in a child with Down's syndrome and transient chromosome aberrations in the mother

We wish to report a trisomy 21 child with two supernumerary fragments, whose mother had possibly related transient chromosome aberrations.

The proband was an 8-year-old boy with typical features of Down's syndrome. The lymphocyte chromosomes studied at the ages of 3.5 months and 8 years showed a mosaic karyotype with both cell lines with trisomy 21 and one line (52% of the cells) with two additional fragments (fig 1). The fragments were different sizes with the smaller one resembling the so-called minute chromosomes. The origin of the markers remains unknown despite staining with different methods (standard Giemsa, G, Q, C, and N bands and Harlequin staining). The larger fragment showed some size variation from cell to cell, suggesting ring formation. The fragments had no satellites and were C and N band negative (fig 1b, c). Harlequin staining revealed that the smaller one replicated earlier than the larger one (fig 1e).

Karyotypes of both parents were normal. In 60 out of 200 cells of the mother, however, major chromosome aberrations such as dicentric and ring chromosomes plus acentric fragments were seen (fig 2). Eight years later no such changes were seen in 200 cells studied.

The presence of an additional fragment is quite rare and to our knowledge the presence of two dissimilar minute marker chromosomes has not been reported before. The origin of the marker chromosomes could not

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FIG 1 Karyotype abnormalities in the patient with Down's syndrome. The large arrow indicates trisomy 21 and the small ones chromosome fragments. G, C, and N banding as well as Harlequin staining (a-e respectively).

FIG 2 Transient chromosome aberrations in the mother. Arrows indicate a dicentric chromosome, rings, and fragments.

be solved. The phenotypic role of the patient's abnormal karyotype remained unsolved also; the boy had typical Down's syndrome, apparently resulting from trisomy 21.

The additional fragments must have arisen either as new mutations or have been inherited from one of the parents. In this case, the chromosome complements of the parents were normal despite the transient chromosome aberrations of the mother. Also, the mosaic nature of the patient's karyotype suggests a new mutation.

One possible interesting explanation would be that the mechanism causing the aberrations in the mother could also have caused chromosome fragmentation in the postzygotic cell in the embryo. It has been well documented in various animal experiments that several mutagenic agents can cause congenital chromosome aberrations.¹ In our case no indication was found that the mother had been exposed to infection, radiation, or mutagenic chemicals. However, such a possibility should be kept in mind when studying chromosome aberrations in man.

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