

References

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Long arm deletion of chromosome 22

We report the case of a 13 year old male with a seizure disorder and developmental delay, who has a deletion of chromosome 22 in all peripheral blood lymphocytes examined. To our knowledge, this particular chromosome abnormality has not been previously identified.

A deletion of a G group chromosome was first observed in an infant in 1964.¹ In 1968 it was suggested that there were two distinct syndromes, one in which the physical features were caused by an abnormality of chromosome 21 (G deletion syndrome I) and the other in which an abnormality of chromosome 22 was the cause of the abnormal physical features (G deletion syndrome II). The characteristics associated with G deletion syndrome I are: mental retardation, hypertonia, microcephaly, large or low set ears, skeletal malformations, and growth retardation. The characteristics associated with G deletion syndrome II are: mental retardation, hypotonia, epicanthic folds, and syndactyly of the toes.²

These chromosome abnormalities and their associated physical characteristics are somewhat variable, as noted by Maeda *et al.*³ They concluded that the physical features associated with G deletion syndrome I are constant and represent a clinically distinct syndrome. The physical characteristics associated with G deletion syndrome II, on the other hand, appear to be somewhat variable.

We describe here the clinical and cytogenetic findings in a 13 year old male who has a deletion of the long arm of chromosome 22. The patient was born after a 10 month pregnancy to a deaf, 18 year old, G1P0 woman and a deaf 17 year old man. The child sat alone at 12 months, crawled at 18 months, and walked at three years. He frequently exhibits physically aggressive behaviour. The child has poor communication skills that cannot be attributed to his parents' lack of speech. Auditory sensitivity for simple speech and tonal stimuli were assessed and hearing impairment as a contributing factor to his speech delay was ruled out. Other areas of development, including gross and fine motor development, self help skills development, and cognitive development were also delayed. Physical exami-

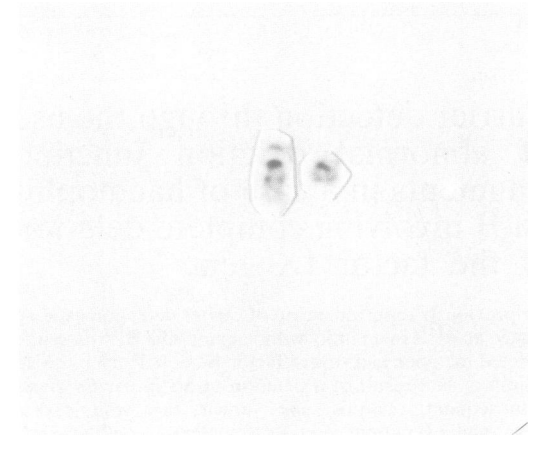


FIGURE G banded example of 22q—.

nation revealed bilateral epicanthic folds, as well as broad fingers and thumbs. No other external abnormalities were noted.

All metaphases examined (122) showed a 46,XY,del(22)(pter→q12:) complement (figure). The chromosomes of both parents were normal.

The clinical features found in our patient with a deletion of chromosome 22 do not coincide completely with those observed in G deletion syndrome II. While our patient is mentally retarded and has epicanthic folds, he does not have syndactyly of the toes or hypotonia, both of which were observed in all cases of G deletion syndrome II. In fact, our patient exhibited retarded psychomotor development which is associated with G deletion syndrome I. This is consistent with the variability reported in G deletion syndrome II by other authors. A compilation of additional cases is necessary to clarify the syndrome associated with deletion of the long arm of chromosome 22.

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