

Case reports

Partial monosomy 8p with minimal dysmorphic signs

E Blennow, K Brøndum-Nielsen

Abstract

A female patient with a 46,XX,del(8)(p23→pter) karyotype is presented. She was mentally retarded and showed a few dysmorphic features. Her red cell glutathione reductase level was within normal limits. This terminal deletion, on the short arm of chromosome 8, appears to be the smallest segment hitherto reported.

Since 1973 11 children have been reported with a partial deletion of 8p. Further cases have had various unbalanced translocations with partial trisomies in addition to monosomy 8p. Some of the features commonly described are mental retardation, growth retardation, a high, narrow forehead, epicanthic folds, short neck, wide set nipples, congenital heart defects, and, in males, genital abnormalities. In one case the level of glutathione reductase was reduced. This locus has a confirmed assignment to p21.1.¹

We describe a case whose appearance is not obviously dysmorphic and whose main features are mental retardation and behavioural problems.

Case report

The female proband was born in 1972 and was the first child of a 28 year old woman and a 30 year old man. Pregnancy and delivery were uneventful. Birth weight was 3430 g and length 50.5 cm. At birth physical examination showed convergent strabismus. She was otherwise considered normal although, according to the mother, she suffered from episodes of shakiness.

She did not walk until 20 months of age. As she grew older the mother found that she was slow in

development. Examination at 3 years 9 months showed an overall delay in psychomotor development, but social skills were most delayed (about two years). She was indiscriminate in her contact, could not concentrate, and was easily distracted. At the age of 7 the proband was admitted to a special school. Puberty was normal with menarche at 13 years. Mental retardation was within the moderate range, but according to her parents her memory was remarkable and she speaks two languages (her parents originate from Germany). She was hyperactive.

On physical examination at the age of 16, her appearance was not obviously dysmorphic although her facial features were somewhat coarse with a prominent nose (fig 1). The palpebral fissures had an antimongoloid slant and slight hypotelorism was present. A convergent strabismus was noted. The forehead was narrow and the chin slightly receding. The ears were normal. Her hands were small and slender. The height was 163 cm, weight 61 kg, and head circumference 55 cm. Her body proportions

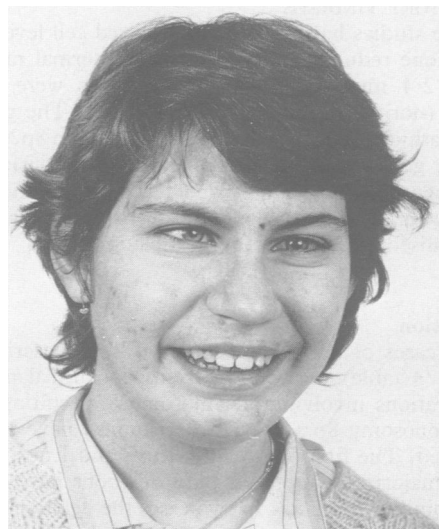


Figure 1 The proband at 14 years.

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paucity of aberrant phenotypic features. Except for two boys described by Taillemite *et al*³ and Bresson *et al*,⁶ all these children have shown markedly retarded postnatal growth, and they have all, with one exception,⁴ been small for dates at birth. The clinical picture has varied, but most of them have only had minor dysmorphic features, including epicanthic folds, ear anomalies, and wide set nipples. Other features are short neck, retrognathia, and a narrow, high forehead. Congenital heart defects are common and so are genital abnormalities in the males. All these signs are rather unspecific and the main feature is psychomotor retardation. However, some authors consider these features to constitute a recognisable syndrome.^{4 8-12}

Our case was neither small for dates nor did she show growth retardation. She had very few dysmorphic signs but behavioural problems were present, as reported in the case of Rodewald *et al*.⁸ The breakpoint of the deletion was more distal than in any of the previously reported cases, and the red cell level of glutathione reductase was normal in our patient, which also indicates that the deletion was terminal rather than interstitial.

The clinical picture of 8p- syndrome seems to show distinct features. The combination of retarded somatic growth, mental retardation, and minor physical abnormalities, including slightly dysmorphic

facial features, may suggest a chromosomal aberration, and high resolution banding is indicated in such cases.

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