The data for propranolol were also analysed: 0·12% and 0·19% of mothers of matched healthy newborns and total malformed index patients, respectively, were treated (table 2). This figure was somewhat higher in the groups of cleft lip/cleft palate, neural tube defect, and hypospadias. However, a comparison with their matched controls did not indicate any significant difference.

Our database does not contain a pregnant woman who was treated with both ergotamine and propranolol during pregnancy.

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References

18p− syndrome with partial sacral agenesis
Sir,

We should like to present additional information regarding a patient with 18p− and a single central maxillary incisor who was originally reported in 1981.1 The patient, now aged seven years, was initially seen for evaluation of short stature, developmental delay, and unusual facial features, most notably a single central maxillary incisor. Growth hormone studies were unremarkable, but a lymphocyte karyotype showed 18p− in all cells examined. Karyotypes of the parents were normal.1

Toilet training was attempted but was not successful; this lack of success was ascribed to the patient’s developmental delay. Cognitive skills were in the range of borderline normal intelligence to mild mental retardation. A barium enema and intravenous pyelogram at the age of four years were reported to be normal, although spina bifida occulta (level not specified) was noted. Encopresis and enuresis persisted at the age of seven years. Because of the previously normal barium study and the normal neurological examination, it was felt that the encopresis and enuresis did not have an organic aetiology and a behavioural modification programme was instituted. This was not successful and, on further study, a radiograph of the lumbosacral spine showed partial agenesis of the sacrum. This was confirmed by a CT scan and myelogram of the sacrum, which also showed a high termination of the theca (at L4) and spinal cord (at T12 and L1), with no evidence of an intraspinal mass lesion.

Partial sacral agenesis with spinal cord abnormalities has not been described in the 18p− syndrome as far as we are aware. The neurological dysfunction
Correspondence

FIGURE. Patient at the age of seven years. Note the central maxillary incisor, prominent ears, and fleshy nasal tip.

which has apparently resulted from this malformation has caused considerable problems for this child in terms of enuresis and encopresis, without obvious neurological deficits in the legs. Bowel and bladder problems are not unusual in children with developmental delay or mental retardation, and it is tempting to associate these problems with poor intellect, behaviour, or difficulties with parenting. The possibility of a structural basis for these problems should be considered, particularly in a child with other malformations or a chromosomal abnormality. The stance abnormalities associated with 18p−2 might also be related to sacral anomalies, although this possibility has not been investigated in most reported patients.

It is noteworthy that the single central maxillary incisor, first noted in the primary dentition, has also occurred in the permanent dentition (figure).

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References


Some notes for contributors on nomenclature

Nomenclature. Authors should refer to the following publications.


18p− syndrome with partial sacral agenesis.

S Anderson-Shotwell and W G Wilson

*J Med Genet* 1989 26: 70-71
doi: 10.1136/jmg.26.1.70

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