Case reports


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Cat eye syndrome associated with aganglionosis of the small and large intestine


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SUMMARY A newborn male infant is presented with the characteristic phenotype of the cat eye syndrome and a small supernumerary chromosome shorter than a 22. He also had complete absence of parasympathetic ganglion cells throughout the small and large intestine.

The cat eye syndrome is characterised by anal atresia, ocular coloboma, cardiac defects, preauricular tags or sinuses, abnormalities of the urinary tract, mental retardation, and a small supernumerary, bisatellited, isodicentric chromosome. Molecular hybridisation with chromosome 22 specific probes have shown that the isodicentric chromosome

FIG 1 Coloboma of the iris and preauricular pits.
A single maxillary incisor as a manifestation of an ectodermal dysplasia

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SUMMARY A single, central, maxillary incisor was found in a patient with an ectodermal dysplasia.

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absence of parasympathetic ganglion cells. The child died at three months of age; this was attributed to sepsis related to a central catheter for parenteral hyperalimentation.

CYTOGENETIC STUDIES
Cytogenetic studies on peripheral blood cells of the patient showed 47 chromosomes in all cells, with a supernumerary chromosome smaller than a G chromosome (fig 2). The karyotype of both parents was normal.

Discussion
Our diagnosis was made according to the minimal clinical criteria of Hsu and Hirschhorn, in that our patient had a combination of two major features, coloboma of the iris and anal atresia, plus one of the most frequent associated anomalies, preauricular pits. Also, the cytogenetic results in lymphocytes using G banding showed a small supernumerary chromosome shorter than a 22.

The outstanding feature in this case is the absence of parasympathetic ganglion cells throughout the intestinal tract. To our knowledge this is the first case of cat eye syndrome associated with a complete absence of ganglion cells in the large and small intestine. However, this association may be coincidental.

References

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