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.

Clinical geneticists continue to encounter new types of ectodermal dysplasia each with its own mode of inheritance. The problem is that precise identification is essential for appropriate genetic counselling. We present two sibs with an abnormality of hair, nails, and teeth in which the dental anomaly in one of the