The Pallister-Hall syndrome

In their report of sibs with the Pallister-Hall syndrome, Thomas et al. make the points that choanal atresia may be a feature of this condition and that it may be caused by a dominant gene. We have recently encountered an infant in whom the findings lend support to both of these suggestions.

Our patient, a female, was the first child born to a healthy 31 year old female and her healthy and unrelated 38 year old partner, both of whom had three healthy children from previous relationships. Birth weight was 3780 g at term. Active resuscitation was needed and during the first day of life she required both ventilator support and intravenous glucose because of hypoglycaemia. Examination showed an unusual facial appearance with a flat nasal bridge, short nose, and anteverted nares. Other findings included a small exomphalos and postaxial polydactyly involving the left hand in which the fourth metacarpal was severely hypoplastic.

Cranial CT scan showed a large, non-enhancing suprasellar mass with a posterior cystic component (fig 1). The clinical impression of osseous bilateral choanal atresia was confirmed (fig 2). Abdominal ultrasound showed mild bilateral renal hypoplasia. Chromosomal examination showed a normal female karyotype.

Subsequent investigation showed evidence of panhypopituitarism and appropriate hormonal and electrolyte supplementation was initiated. The child continued to be ventilator dependent until bilateral surgical correction of the choanal atresia was undertaken at 16 days, after which her respiratory condition improved considerably. However, she continued to be extremely ill with several episodes of bradycardia culminating in death at the age of 48 days. Permission for necropsy was withheld.

The clinical features in this child are strongly suggestive of the Pallister-Hall syndrome, in which choanal atresia appears to be an uncommon but potentially very important finding. No abnormality was present in either parent. The advanced paternal age of 38 years is consistent with a new dominant mutation. Taking into account the paucity of parental consanguinity in the published cases along with the findings in the father of the sibs described by Thomas et al, we agree that it is premature to conclude that the Pallister-Hall syndrome shows autosomal recessive inheritance.

Familial Pallister-Hall syndrome

We read with great interest the report by Thomas et al. describing two sibs affected with Pallister-Hall syndrome (PHS) and a father who manifests polydactyly. We have reported a family with male to male transmission of PHS and also concluded that many, if not all, cases of PHS are inherited in an autosomal dominant pattern with variable expression or gonadal mosaicism accounting for phenotypically normal parents. Two reports describing affected sibs with apparently unaffected parents have been published subsequent to the submission of the manuscript by Thomas et al.

A surprising and revealing aspect of the family described in our report was the fact that the father had no apparent neurological or endocrinological symptoms in spite of the presence of a large tumour with putative dysplasia. The report by Thomas et al. does not mention if the father was examined for the presence of an asymptomatic hamartoma. We would recommend that such an examination be considered in this case for the reasons outlined in our report.

The increasing number of persons identified with familial PHS underscores the point made by Thomas et al. that genetic counselling in PHS should be performed with great caution. In addition, the notion that PHS is a uniformly lethal disorder must be discarded.

Dr John Graham of Cedars-Sinai Los Angeles and Dr Biesecker have initiated a collaborative research effort to collect information on all patients with PHS in order to clarify the range of the phenotype of this disorder and to work towards characterisation of the molecular defect in PHS. If the readers of the Journal are aware of any cases of PHS we would be most grateful for any information that is available.


The Pallister-Hall syndrome.

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