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Porencephalic cyst in pycnodysostosis

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SUMMARY We describe a case of pycnodysostosis with porencephaly and suggest an explanation for the porencephaly by a mechanism of imbalance between brain growth and its vascular supply and a normal but unopposed cerebrospinal fluid pressure.

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In 1962 Maroteaux and Lamy¹ described a skeletal dysplasia the main features of which are generalised osteosclerosis and moderate dwarfism. They named it pycnodysostosis meaning 'thick bones'.

Other features of pycnodysostosis include underdevelopment of the cranial sinuses; wormian bones; delayed closure of the fontanelles with wide, unfused cranial sutures; a beaked nose; a high, grooved palate; multiple dental abnormalities; hypoplastic

FIG 1 Proband's face with frontal bossing, large beaked nose, and small mandible.

clavicles; acro-osteolysis; and sometimes multiple fractures and mental retardation.²⁻⁷

Case report

The proband, the oldest of two brothers with pycnodysostosis, was examined at 24 years of age. Pregnancy and delivery had apparently been normal. Examination showed short stature (143 cm, below the 3rd centile), an occipitofrontal circumference on the 50th centile (57 cm), frontal bossing, a small mandible, and deep nasolabial skin folds (fig 1). The fontanelles were open and the sutures widely separated. The palate had a submucous cleft and dental dysplasia was apparent. The hands were short (length less than the 3rd centile) with very small

distal phalanges and spoon shaped nails; the skin over the phalanges was wrinkled (fig 2).

The patient had sensorineural deafness and his mother reported "a fracture of his left foot at the age of six years". Developmental milestones seemed to be normal.

Radiological studies showed an abnormal pattern of ossification of the calvarium with open fontanelles, wide sutures with wormian bones, absent frontal sinuses, osteosclerosis of the cranial base, and a small mandible with an obtuse mandibular angle (fig 3). The iliac bones were hypoplastic and osteosclerotic and the bones of the legs and feet were also osteosclerotic. The distal phalanges of both halluces showed acro-osteolysis.

CT scan showed a left frontal porencephalic cyst

FIG 2 Hands of the proband; see text for description.

FIG 3 Cranial x rays of the patient.

FIG 4 CT scan of the proband showing a left frontal porencephalic cyst.

communicating with the ventricular system and covered with a layer of cerebral cortex (fig 4).

The younger brother of the proband showed the same type of bone abnormalities but with a normal CT scan.

Discussion

Some of the skeletal abnormalities present in these patients can be found in other disorders. The differential diagnosis includes cleidocranial dysplasia, Hadju-Cheney syndrome, and mandibuloacral dysplasia.⁷ All may show acro-osteolysis, hypoplastic clavicles, and delayed closure of the fontanelles with large sutures, but without signs of osteosclerosis. In Hadju-Cheney syndrome osteoporosis and multiple fractures are frequent and in mandibuloacral dysplasia acro-osteolysis is severe and osteosclerosis is absent.⁸ In cleidocranial dysplasia bone density is normal and inheritance is autosomal dominant. In our family dominant inheritance was excluded by clinical and radiological examination of both parents. Consanguinity between parents of patients with pycnodysostosis has been reported in about 30% of cases.^{6,8} Its absence in our family does not rule out the existence of hidden endogamy, since both parents were born in the same small village in Madeira.

To our knowledge this is the first report of an intracranial, supratentorial cyst in pycnodysostosis. Porencephaly is a term now used for any cavitation

or CSF filled cyst in the brain.^{9,10} Type I, or encephaloclastic porencephaly, is usually unilateral and results from a destructive lesion of normal brain, such as fetal vascular occlusion (around the fifth month of gestation) or birth trauma.¹¹ Type II, or schizencephalic porencephaly, is usually symmetrical and bilateral and may represent a primary defect in neuroectoderm.¹² Our case is in keeping with porencephaly type I.

We can speculate that abnormal development of the cranial bones with very large and underossified sutures could explain a faulty anatomical relationship between brain growth and its vascular supply. Since in pycnodysostosis the cranial vault could not limit brain growth (leading to a disproportionately large head circumference in relation to length), a normal intraventricular pressure acting unopposed might cause an increased surface of the ventricle walls. Beyond certain limits, normal vascular supply would be insufficient to nourish a progressively distended brain, explaining ischaemic necrosis and cavitation.

We recommend the use of CT scan of the brain as part of the routine examination of pycnodysostosis in order to assess correctly the incidence of structural brain abnormalities in this disorder and as contribution to the understanding of the development of the fetal nervous system.

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