Alobar holoprosencephaly and otocephaly in a female infant with a normal karyotype and placental villitis

The proband was born to a 24-year-old gravida 1, para 0 black woman after a 35 week gestation complicated by polyhydramnios. Labour was induced when ultrasound studies indicated severe congenital malformations. The family and maternal histories, including exposure to viral and toxic agents and developmental disturbances, were negative. The 1280 g premature cyclopic female infant measured 25 cm crown-rump and 40 cm crown-heel (figure a, b).

The major portion of the face was occupied by a proboscis with a midline probe patent ostium. The ostium ended blindly in a space 1 cm in diameter contained within the soft tissue. Bulging from the scalp above the proboscis was an anterior meningocele. Below the proboscis, the external ears were fused and the auditory canals were absent with apparent agenesis of the middle and inner ears. The mandible was absent and there was no oral opening although there was an oropharyngeal space that contained a hypoplastic tongue. There was a tracheo-oesophageal fistula; the proximal segment of the oesophagus ended as a fibrotic cord and the distal segment was in continuity with the trachea, entering just above the carina. The brain was holoprosencephalic, alobar type, with a single large ventricle and fused thalami. The central cerebral mass consisted of grey matter and ependymal lined spaces. Cranial nerves I, II, III, and IV were absent. The remainder of the central nervous system appeared within normal limits. The pituitary gland was not identified and the adrenal glands were hypoplastic.

There were many cardiopulmonary anomalies. The configuration of the heart was that of a double outlet right ventricle with subaortic ventricular septal defect and complete pulmonary stenosis. The right atrium was dilated and the right ventricle hypertrophied. The foramen ovale and ductus arteriosus were patent. Only the left coronary artery was present. The right pulmonary artery and vein were atretic and the right lung was half the size of the left.

The placenta weighed 344 g and microscopically exhibited multifocal areas of chronic necrotising and granulomatous villitis, haemorrhagic villitis and endovasculitis, and chronic granulomatous chorionitis.

The karyotype of the proband was normal, 46,XX.

The patient is an example of alobar holoprosencephaly combined with otocephaly, a combination of anomalies that is reported infrequently. It is noteworthy that in this case the karyotype was normal. The placental changes were suggestive of intrauterine infection, but while haemorrhagic endovasculitis and villitis have been associated with toxoplasma, rubella, cytomegalovirus, and herpes infections, these were not found in this patient.

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References

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FIGURE (a) Face of proband. (b) Face of proband with head extended showing otocephalic deformity.

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