Congenital absence of the nose: a case report and literature review

Abstract A case of congenital absence of the nose is presented. The etiology of this rare condition is unknown. A review of the literature reveals that the previously applied terms, e.g. ‘arhinia’, are unclear. In the reviewed cases there seems to be a pattern of facial anomalies associated with nasal absence. In most cases, one could probably expect a lack of the olfactory bulbs and tracts. We suggest a new terminology and summarize the aims of the radiological evaluation of this condition.

Introduction

Congenital absence of the nose is rare. A review of 22 previously reported cases of congenital absence of nasal cavities and external nose, but without holoprosencephaly, is presented to illustrate the complexity of the anomaly. We also report on a new case, which was recognized by prenatal ultrasound after 17 weeks’ gestation and further investigated with CT and MRI after birth.

Several classification systems have been proposed for these conditions [1–3], but the previously used terms ‘arhinia’, ‘arhinencephaly’ and ‘arhinogenesia’ are still not clearly defined. Cases of familial absence of the nose have been reported [4, 5], but the contributions of genetic as well as maternal factors in these malformations remain unknown.

Case report

Our case was a white female. She was the first child of healthy and unrelated parents. The mother was not on any medication, and there was no family history of congenital malformations. A routine
US examination in the 17th gestational week demonstrated diffuse midfacial anomalies with oedema, and absence of the external nose was diagnosed in the 25th week (Fig. 1). Tests for TORCH and parvovirus were negative, and the karyotype was normal. The child was delivered by caesarean section after 37 weeks’ gestation due to polyhydramnios and repeated amniocentesis. A two-vessel umbilical cord was found. Birth weight was 3,070 g and APGAR scores were 9 at 1 min and 9 at 5 min. The clinical examination confirmed absence of the external nose. In addition, there was a high arched palate, slight hypertelorism and bilateral coloboma of the iris. The vital signs were otherwise normal.

Shortly after birth the baby was admitted to the neonatal intensive care unit due to slight respiratory distress, which was relieved by an oropharyngeal tube. The baby recovered completely after 2 weeks. There were several readmissions due to recurrent dacrocystitis requiring antibiotic therapy. Her psychomotor development was normal. At the age of 8 months the child was still partly fed via a nasogastric tube. Constructive surgery has been deferred, probably until pre-school age.

Cerebral US (HD1 5000, ATL, 8–5-MHz micro-curved probe) in the neonatal period was normal. Facial CT (High Speed Advantage, GE Medical Systems, 1-mm axial scans, pitch 1.0. Figs. 2, 3) showed absence of nasal bones, cribriform plate and septal structures, including vomer, perpendicular plate of ethmoid and septal cartilage. The frontal processes of the maxillae fused apically under the glabella. The maxillae were hypoplastic with a high arched bony palate. The potential nasal fossae were filled with soft tissue and paranasal sinuses were missing. Cranial MR (Siemens Magnetom Vision, 1.5 T. Figs. 4, 5) confirmed the CT findings. In addition, a non-communicating air-filled cavity was seen between the soft palate and the palatine tonsils, representing a rudimentary nasopharynx. A 10-mm soft-tissue layer in the atretic nasal cavity separated the hard palate from the floor of the anterior cranial fossa. No encephalomeningocele was seen. The olfactory bulbs were missing, and there was no olfactory sulcus, the rectus and medial orbital gyri being fused. A cystic tumour was seen in the place of the left lacrimal duct, representing a mucocele, probably due to nasolacrimal duct atresia.

**Discussion**

The nose has a complex development (Fig. 6). From 4 weeks’ gestation the frontonasal processes appear and slowly elevate into the dorsum and apex of the nose. During the 5th week, the nasal placodes, which are ovoid thickenings of epidermal ectoderm, invaginate, forming the nasal pits. The nasal pits deepen dorsocaudally to form the nasal part of the oronasal cavity. Mesenchymal proliferations in the margins of these pits give rise to the lateral and the medial nasal processes. The paired medial processes subsequently fuse, forming the bridge of the nose, the nasal septum and