Nasopharyngeal teratoma ('hairy polyp'), Dandy-Walker malformation, diaphragmatic hernia, and other anomalies in a female infant

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Abstract

Nasopharyngeal teratomas are rare and are infrequently associated with extra-oral malformations. The case of a premature female infant with multiple congenital anomalies, including nasopharyngeal teratoma, Dandy-Walker malformation, diaphragmatic hernia, and congenital heart defect, is presented. This constellation of malformations does not appear to have been reported previously.

Case report

A female infant was born at 30 weeks of gestation to a 30 year old woman. The pregnancy was complicated by vaginal spotting between the 14th and 19th weeks, and by premature rupture of the membranes; there was no history of exposure to teratogens. Fetal sonography undertaken two days before delivery showed macrocephaly, probable Dandy-Walker malformation, and left diaphragmatic hernia. The patient was delivered vaginally after spontaneous onset of labour. Apgar score was 1 at one and five minutes. The patient died at 2 hours of age.

Birth weight was 1470 g (slightly >-1 SD), length was 40 cm (mean), and OFC was 39 cm ($>>+2$ SD).

Examination showed massive macrocephaly (fig 1) with widely spaced sutures and a broad forehead. The anterior fontanelle was 3 cm in greatest coronal diameter; estimation of its sagittal diameter was precluded by the extreme suture diastasis. The facies (fig 2) was broad, triangular, and flattened, with midfacial hypoplasia, hypertelorism (inner canthal distance 26 mm ($>>+2$ SD), outer canthal distance 55 mm ($>+2$ SD)), downward slanting palpebral fissures (palpebral fissure length 16 mm OD (75th to 90th centile), 14 mm OS (25th centile)), and infra-orbital creases. The nasal bridge was broad and the nares were anteverted. A long, narrow median cleft of the secondary palate was present; through this protruded a large mass, similar in appearance and consistency to tongue, upon which fine hairs were sparsely distributed (fig 3). The tongue was of normal size and had a median ridge posteriorly. The mandible

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Figure 1 Necropsy photograph of the patient showing macrocephaly.

was quite small. The pinnae were 24 mm long bilaterally (25th centile); they were soft, low set, and retroverted with poorly formed helices. The left pinna (fig 2c) was triangular with a prominent and straight antihelix. The abdomen was scaphoid. Anogenital examination was normal. The hands and feet were gracile, without polydactyly, syndactyly, or brachydactyly. The left hand was 45 mm long (slightly < mean); the left middle finger was 18 mm long (-1 SD); and the left foot was 55 mm long (slightly < mean). Mild bilateral talipes valgus was present. The palmar creases were normal. There was a longitudinal crease on the left sole.

Additional findings at necropsy included a fibrous band, which connected the median ridge of the tongue and the left side of the hard palate. Gross examination of the oral mass was consistent with a nasopharyngeal teratoma. The right tympanic membrane was absent. The cerebral gyral pattern was consistent with the gestational age. The cerebellum was hypoplastic. The cerebellar vermis was absent. A 10 cm, thin walled cyst occupied the posterior fossa and passed through a 5 cm tentorial notch. There was agenesis of the corpus callosum with Probst bundles and cavum septi pellucidi et vergi. The ventricles were enlarged and the grey-white demarcation was indistinct. These findings were consistent with Dandy-Walker malformation. A defect of the left hemidiaphragm was present, through which herniated the left and quadrate lobes of the liver, the stomach, the small bowel, and most of the large bowel. The left lung was hypoplastic and the right lung was bilobular. The heart was dextrorotated and displaced into the right pleural cavity. A persistent left superior vena

Figure 3 Frontal view of patient showing hairy polyp (p) and tongue (t).

cava drained into the right atrium, through the coronary sinus, and preductal coarctation of the aorta was present. The bladder musculature was mildly hypertrophic; no urinary tract obstruction was present. The alimentary tract, liver, gall bladder, pancreas, kidneys, uterus, ovaries, and spleen were normal; the thymus weighed 2.5 g (normal = 2.8 to 7.0 g).

Figure 2 Frontal (a), right lateral (b), and left lateral (c) views of the patient.

Histologically, the tongue was normal. The oral band was fibromuscular and was covered with stratified squamous epithelium. The oral mass was covered with keratinised, stratified squamous epithelium, containing numerous hair follicles and sebaceous glands. A portion of the mass resembled primitive neural and mesenchymal tissues. A pathological diagnosis of 'hairy polyp' was made. Routine G banded karyotype was normal (46,XX).

Family history was remarkable for partial cutaneous syndactyly of the hands and feet and an unspecified renal abnormality in the patient's father. The paternal grandmother had had nine first trimester spontaneous abortions, with the patient's father being her only liveborn child. A maternal great grandmother and the spouse of a maternal uncle each had had two spontaneous abortions. A maternal first cousin once removed had Down's syndrome. A maternal half sister was alive and well. There was no known consanguinity.

Discussion

Unlike other teratomas of the head and neck (teratoids, teratomas, and epignathi, each of which contain elements derived from all three germinal layers (endoderm, mesoderm, and ectoderm)), 'hairy polyps' are bigeminal, being derived from mesoderm and ectoderm only. Although hairy polyps are the most common type of head and neck teratoma, they are nevertheless rare; about 60% originate in the nasopharynx, with most of the remainder occurring in the oropharynx. Nasopharyngeal teratomas may interfere with closure of the palatal shelves, and thus are frequently associated with cleft palate.¹ Reports of hairy polyps associated with extraoral anomalies are few,²⁻⁴ and the pattern of malformations seen in our patient appears not to have been recognised previously.

Of the known multiple congenital anomaly syndromes, Fryns syndrome⁵ is most consistent with the facial appearance, cleft palate, Dandy-Walker malformation, cerebellar anomalies, and diaphragmatic hernia seen in our patient; cardiac anomalies may also be seen in that condition. However, our patient had no apparent distal limb hypoplasia, and she did not have bicornuate uterus or renal anomalies. The failure of palatal closure presumably resulted from the intervening oral mass, and thus the pathogenesis of her cleft palate appears to be different from that typical of Fryns syndrome. Finally, nasopharyngeal teratoma has not previously been associated with Fryns syndrome.

Although both the acrocallosal syndrome⁶ and the hydrolethrus syndrome⁷ may be associated with agenesis of the corpus callosum and cleft palate, our patient's features are not otherwise typical of either of these conditions. Specifically, her lack of polydactyly (which is a distinguishing, albeit not absolutely invariable, feature of each of these conditions) strongly militates against the diagnosis of either of these syndromes.

Dandy-Walker malformation and congenital heart defects (CHD) frequently occur together,⁸ as do diaphragmatic hernia and CHD.⁹ Furthermore, epignathus has been associated with central nervous system malformations.¹⁰ However, concurrence of Dandy-Walker malformation, CHD, and hairy polyp, as seen in our patient, is apparently unique.

A relationship between the anomalies of the patient (which did not involve the digits or kidneys) and those of her father is not evident. Furthermore, given the patient's normal chromosomes, the relevance of the family history of recurrent spontaneous pregnancy loss is also uncertain, although the possibility of chromosomal mosaicism or of a submicroscopic chromosome abnormality cannot be excluded.

Thus, although we cannot exclude the possibility that our patient may represent an unusual case of Fryns syndrome, it is more likely that she had instead a unique constellation of congenital anomalies, the most unusual of which is hairy polyp.

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