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AJNR Am J Neuroradiol 1992, 13 (5) 1471-1476

<http://www.ajnr.org/content/13/5/1471>

This information is current as
of April 17, 2024.

Proboscis Lateralis with Associated Orbital Cyst: Detailed MR and CT Imaging and Correlative Embryopathy

Larry B. Poe,¹ Leo Hochhauser,¹ Christine Bryke,² Barbara W. Streeten,³ and Jannell Sloan²

Summary: Proboscis lateralis is a rare craniofacial malformation. We present a case in a 1-week-old male infant, describe the clinical and imaging findings, and discuss the pertinent embryology. MR and CT proved to be complementary: CT provided anatomic detail in bone, defined the nasal cavity, and aided in determining the relationship of the proboscis and orbit; MR clarified the relationship of the proboscis to the orbit and skull base, and the relationships of normal brain to the dysplastic ethmoid centers.

Index terms: Orbits, computed tomography; Orbits, magnetic resonance; Orbits, abnormalities and anomalies

Proboscis lateralis (nasal aplasia with proboscis) is a rare congenital craniofacial anomaly occurring in less than 1/100,000 births. Proboscis is derived from the Greek word "proboskis" for trunk. This soft tubular process typically measures 2–3 cm in length and 1 cm in diameter and arises from the inner canthus of the eye (1). This anomaly has a complex embryology that is important for understanding its development and associated facial defects. Although a few cases have been reported in the literature, our case is unique in having highly detailed correlative imaging anatomy prior to surgical correction.

Subject and Methods

B.L. was the 9 lb 3 1/2 oz product of a full-term gestation of a 21-year-old G1P1 white woman who delivered by normal spontaneous vaginal delivery. Prenatal history was negative for teratogenic exposure. There was no family history of birth defects. The parents were not consanguineous.

Physical examination at 8 days of life revealed a vigorous, well-proportioned, large-for-age white male infant with dysmorphic features limited to the midface (Fig. 1). Most prominent was a tubular structure, 3.0 × 1.5 cm, projecting outward and downward from its origin from medial to the

left inner canthus. A clear mucous secretion exuded from its distal orifice when the infant cried. The left side of the nose was aplastic. Just inferior to the nasal septum was a 2-mm wide opening in the upper philtrum. The left palpebral fissure and globe were smaller than the right and a cleft was present at the left inner canthus. The left red reflex was weaker than the right. The remainder of the baby's examination, including the neurologic examination, was unremarkable. Visual-evoked response testing revealed no potential on the left and a poorly formed and delayed potential on the right.

This child underwent computed tomography (CT) at 8 days of age without iodinated contrast enhancement on a GE 9800 scanner utilizing 1.5-mm sections through the face in axial and coronal planes. Three-dimensional computer-generated reconstruction was performed utilizing 3-D Quick software. Subsequently, at 16 days of age, magnetic resonance (MR) imaging was performed without gadolinium DTPA contrast enhancement on a GE 1.5-T Signa system. Axial, coronal, and sagittal planes were imaged utilizing 3- to 5-mm thick sections and TR/TE/excitations ranging from 600-2200/17-90/1-2.

Results

Imaging Findings

Axial and coronal CT scans (1.5 mm) with 3-D reconstructions (Figs. 2 and 3) and MR imaging in three planes (Figs. 4 and 5) demonstrated the trunk-like proboscis emanating from the medial canthus of the left eye. The proboscis was completely extraorbital and was composed of various tissues, including bone within its walls. It had a pneumatized central canal lined by soft tissue that did not communicate with the nasopharynx. The single right-sided nasal passage did communicate with the nasopharynx. The ethmoid air cells were only developed on the right side. There was absence of a definite crista galli. The fovea

Received July 16, 1991; accepted and revision requested October 22; revision received December 4.

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AJNR 13:1471-1476, Sep/Oct 1992 0195-6108/92/1305-1471 © American Society of Neuroradiology

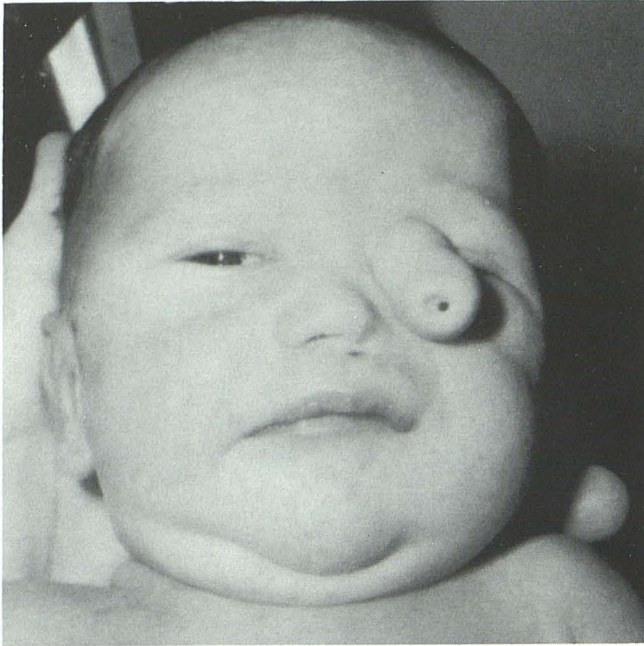


Fig. 1. Photograph of the 8-day-old infant showing left nasal aplasia and a tubular proboscis measuring 3×1.5 cm in length arising just medial to the inner canthus of the left eye. Orifice of a 2-mm blind-ended pit is present in the upper philtrum just inferior to the nasal septum.

ethmoidalis and cribriform plate were not evident; however, incomplete ossification of the skull base in this location is expected in newborns (2). Although the left gyrus rectus protruded more caudally (Fig. 5) than the opposite side, no definite extension through the expected region of the foramen cecum or fovea ethmoidalis was identified. A cephalocele was not expected and was not found at surgery. The turbinates were unilaterally absent on the left. The left orbit and globe were small. A large retrobulbar mass was present within the orbit that either completely obscured or replaced the optic nerve and was presumed to be "cystic degeneration of the nerve," as previously described by Wang et al. (3), although a congenital tumor such as a lymphangioma, glioma, or even sarcoma could not be excluded. A follow-up MR performed 2 months later substantiated the benign nature of the orbital mass. A small bony defect in the floor of the left anterior cranial fossa was discovered preoperatively and substantiated at surgery.

MR and CT complemented each other. CT provided the details of the bony anatomy, including the truly ossified portions of the anterior skull base versus dura or fibrous tissue that were also dark on MR; defined better details of the nasal cavity; aided in determining the relationship of

the proboscis and the orbit; and showed that a portion of the proboscis wall was ossified. MR clarified the relationship of the proboscis to the orbit and skull base and demonstrated the relationships of normal brain (and its lack of gross structural anomalies) to the dysplastic ethmoid centers.

Discussion

A brief review of the embryologic development of the face (Fig. 6) is required to understand the etiology of this defect and its relationship to other craniofacial malformations (2, 4–6, 7).

The development of the face is a very complex sequence of events requiring interaction of several primordial fields. A field is that part of the embryo in which control of development of a complex structure has an orderly progression in space and time of differentiated structures (8).

The very early embryo is composed of totipotent cells that are acted on by organizers within a field volume. These organizers have their greatest effect at the center of the field and lesser effects at its periphery. The most important organizer in the head is the notochordal process. By its action, the neural tube is normally closed by 4 weeks of gestation and the paired nasal fields and optic fields emerge. Lateral mesenchyme created by "dropped" cells of the neural crests migrate around the neural tube rostrally to form the unpaired central frontonasal process (6, 9). At 4.5 weeks, bilateral nasal (olfactory) placodes develop by induction of surface ectoderm by the primitive forebrain (prosencephalon) at the rostral end of the embryo. The region between the placodes is referred to as the "interplacode area." As the prosencephalon organizes the formation of the optic and olfactory placodes, the foregut organizes the first branchial arch into a maxillary process. Between 5–6 weeks of gestation, there is transformation of the nasal placodes into pits and then nasal grooves that eventually become primitive nares and blind nasal passages. The nasal grooves along with the frontonasal process induce the formation of three paired facial swellings: the maxillary processes and the medial and lateral nasal processes. The nasal (olfactory) processes eventually induce the olfactory nerves and cribriform plate. The medial nasal process gives rise to a portion of the nasal floor (vomer), philtrum of the upper lip and the premaxilla (primary palate), and columella (bulbus) of the nose. It probably acts along with the

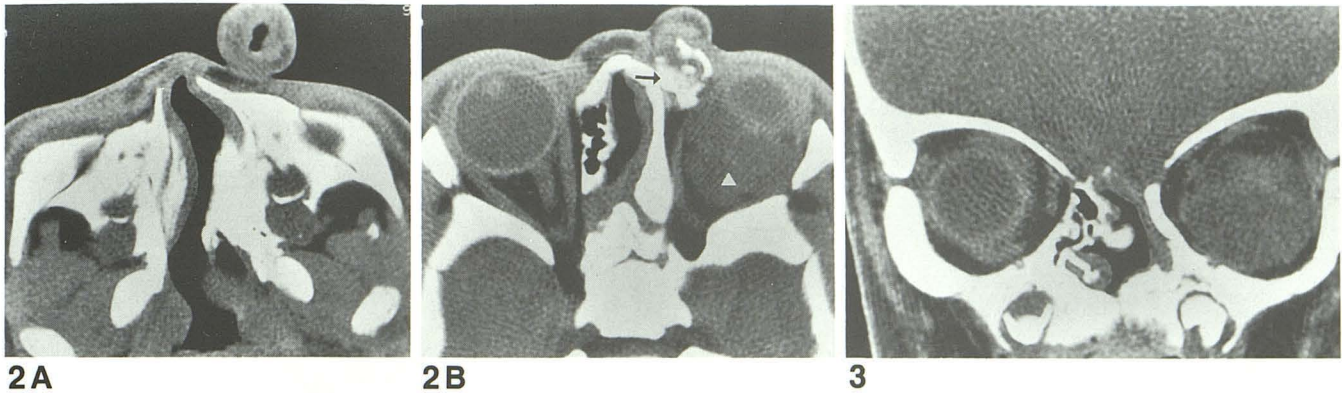


Fig. 2. Axial 1.5-mm noncontrast CT. Soft-tissue windows.

A, Reveals a portion of the proboscis on the left with partially pneumatized central canal. A single nasal passage is present communicating through a single choana with the nasopharynx.

B, A more superior section continues to reveal a single nasal passage. No ethmoid air cells have formed on the left as they have on the right. Ossification within the wall of the proboscis is noted at its attachment to the medial canthus of the orbit (*arrow*). The left eye is microphthalmic. There is a large low-density retrobulbar intraconal mass (*arrowhead*). The optic nerve is obliterated.

Fig. 3. Coronal 1.5-mm noncontrast CT. Soft-tissue windows. The nasal septum is absent. There is a single nasal passage. The turbinates on the left have failed to develop. A large left orbital mass is evident. There is absence of the cribriform plate, left ethmoid sinuses, and ossified left ethmoid roof (*fovea ethmoidalis*) separating the frontal lobe from the nasal passage.

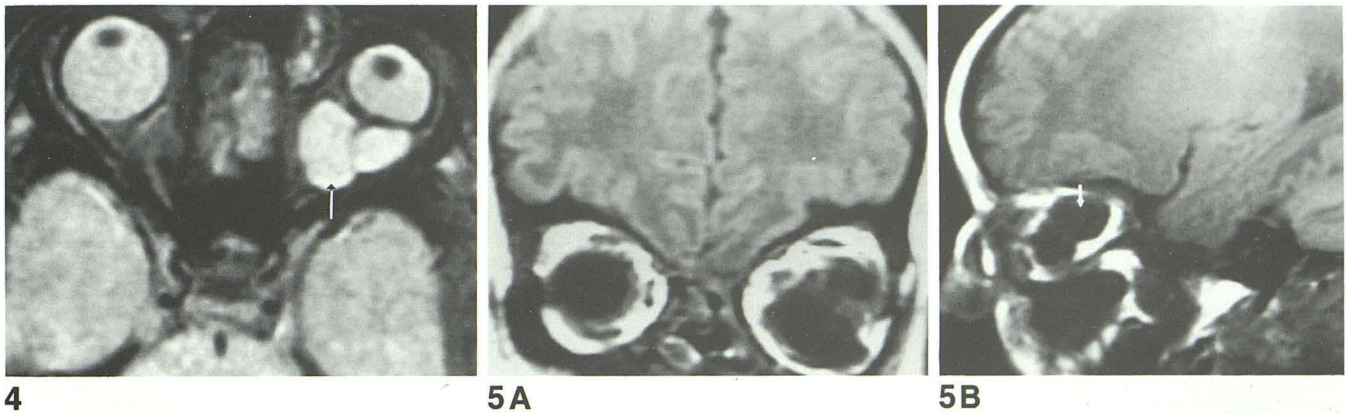


Fig. 4. Axial MR, 2200/90/1 (TR/TE/excitations). These reveal a well-circumscribed left retrobulbar mass that has signal intensity slightly greater than bulk water on the T2-weighted images. The left orbit and globe are small. Soft tissue within the proboscis at the medial canthus is obvious. There is chemical shift artifact within the orbital mass posteriorly (*arrow*).

Fig. 5. Coronal MR (600/17/2) and parasagittal MR (700/17/2).

A, A definite delimiting plate between the left frontal lobe and nasal passage is not present. The gyrus rectus does not extend more caudally than expected to indicate an encephalocele.

B, This reveals the constituents of the protruding proboscis as high-signal fat, intermediate-signal connective tissue and epithelium, and low-signal bone. The low-signal intraconal mass replaces the optic nerve (*arrow*).

interplacode area of the frontonasal process to create the nasal septum. The lateral nasal process produces the nasal bones and the upper and lateral nasal cartilages and nasal ala. Thickened ectoderm on the lateral side of the face is noticeable at 4–5 weeks as the lens placode that will later merge with the optic nerve.

The maxillary process is the key to facial development from this point on. At 6–7 weeks, the maxillary process causes the transformation of the oral cavity. Between 7–8.5 weeks, the eyes and nasal primordia migrates to a more midline

location and the nasal septum differentiates. The maxillary process and its derivatives both influence and are influenced by the optic, otic, and olfactory centers. Therefore, it is very common to find deficient maxillary development with other facial anomalies. Between 7–20 weeks, progressive differentiation of premaxilla (palate), maxilla, and medial and lateral nasal structures continues. Grooves that are formed between adjacent processes normally fuse during this time. The unfused grooves, such as the nasolacrimal groove, define various sites of facial pathologic clefts. By the

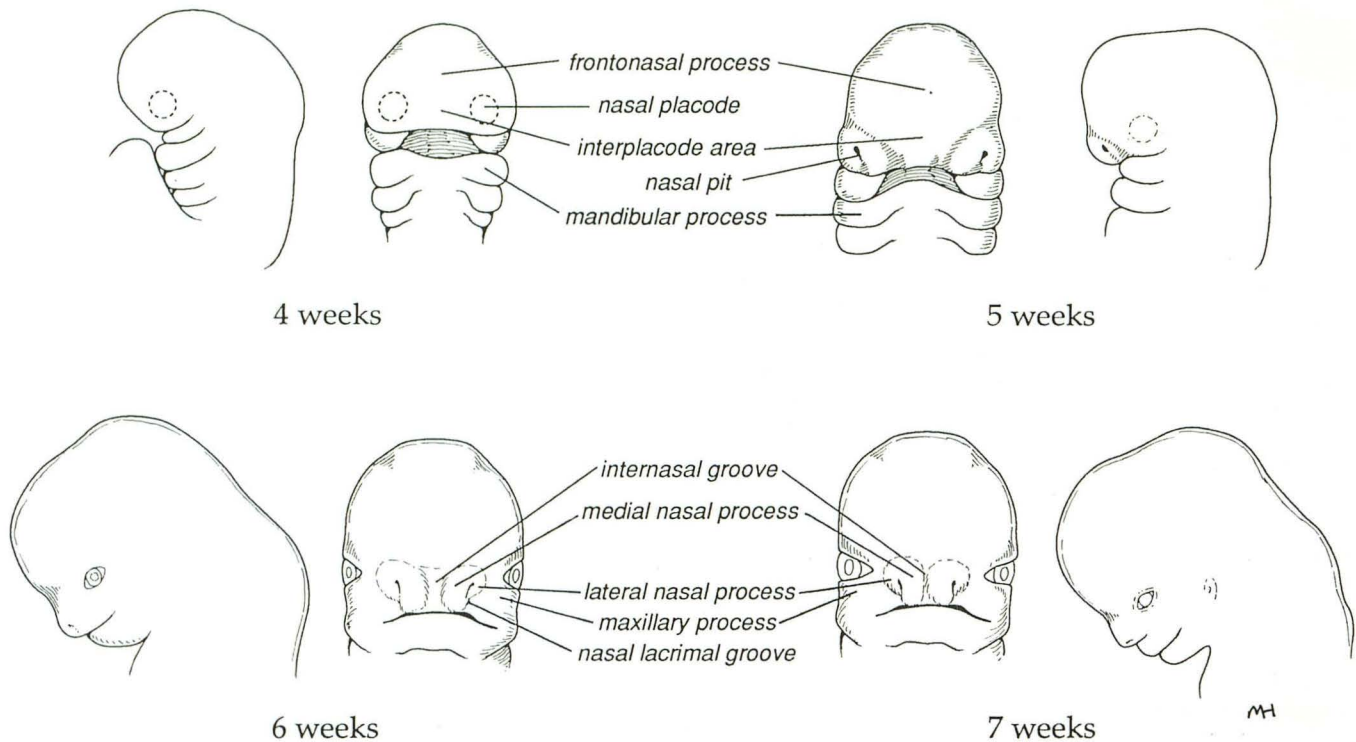


Fig. 6. Diagrams illustrating the embryologic development of the features of the face as it relates to the nose in humans. (Martha Hefner, illustrator)

end of the second month, the nose has a definite appearance. The external nares are plugged by epithelium and are recanalized by 24 weeks. The maxillary process fuses with the lateral nasal process before the medial nasal process fuses. Lack of fusion leads to various forms of facial clefts, as have been extensively reviewed by van der Meulen et al (10) and Tissner (11). The nasolacrimal ducts form via cell canalization. The lower eye lid medially is formed by fusion of mesenchyme along the optic vesicle and maxillary process. The lateral nasal process also contributes to the formation of the medial wall of the orbit. Defects here include a cleft or coloboma of the canthus, iris, retina, or optic nerve. Failure of maxillary growth may lead to hypoplasia of the midface, bifid nose, proboscis lateralis, cleft lip and/or palate, and hypertelorism (6).

A central proboscis may be found in holoprosencephaly. In this severe anomaly, the forebrain fails to diverticulate properly, leading to incompletely divided cerebral hemispheres and ventricles. There is improper formation of the precursors of the frontonasal process (interplacode area). This leads to various states of failure of differentiation of the nasal placodes, resulting in a spectrum of midface anomalies, including cyclopia, cebocephaly, central proboscis, midfacial

clefts and severe hypertelorism, and a central midline proboscis (12).

In our patient at surgery, a frontal craniotomy was performed leading to the discovery of unilateral left-sided aplasia of the olfactory bulb and nerve and partial absence of the crista galli. The aplasia of the olfactory bulb and nerve are designated arrhinencephaly. Although arrhinencephaly may be found in the holoprosencephaly spectrum, it does not in itself constitute a form of holoprosencephaly. In our case, there was no evidence for cerebral midline anomalies.

The exact etiology of proboscis lateralis is unknown (1, 9). Proboscis lateralis (nasal aplasia with proboscis) is a rare congenital craniofacial anomaly occurring in less than 1/100,000 births. As can be surmised above, there is a complex series of events that lead to the development of this malformation. This condition should not be confused with that of central proboscis, which is created by a separate embryopathology in conjunction with the severe cerebral malformation of holoprosencephaly. The embryopathology in proboscis lateralis appears to involve the primary organizers of the nasal portion of the face: namely, the nasal placode and/or forebrain that induces the nasal (olfactory) placode. The inciting event leading to this lesion seems to occur at or

before 5 weeks gestation. The nasal grooves fail to develop properly, but continue to act on mesenchyme, allowing some differentiation into mature nasal tissues such as secretory epithelium and connective tissue. The proboscis itself apparently is derived from the lateral nasal process (1). The primitive mesenchyme grows and fuses in a tubular ring. This soft tubular process typically measures 2-3 cm in length and 1 cm in diameter and arises from the inner canthus of the orbit. The proboscis contains a tiny canal that longitudinally traverses the tube. The canal is lined by nasal columnar epithelium that may secrete mucus. The walls of the tube are composed of mixed connective tissue containing muscle and cartilage and sometimes bone. There is (hemi-) nasal aplasia. The cribriform plate is deficient; the olfactory nerve and nasolacrimal duct absent. The maxillary process fails to induce medial migration of the nasal pits and optic vesicles with resultant hypertelorism. The maxillary contribution to the lateral nasal wall is missing and, therefore, typically there are deficient sinuses, turbinates, and a variety of orbital colobomata (2, 7, 9, 13, 14).

Ophthalmologic defects such as anophthalmia, microphthalmia, and iris, retinal, and lid colobomas are reported to occur in 44% of individuals with proboscis lateralis (1, 6). Boo-Chai's review of the literature uncovered defects in 24 of 34 cases (13). Other facial defects such as maxillary hypoplasia or facial clefts are reportedly seen in 38% (1, 6), with only eight of 34 cases exhibiting these anomalies in Boo-Chai's review (13). Brain and cranial vault disturbances such as encephaloceles or holoprosencephaly are found in 19% (1, 6). Very rarely, there may be an isolated proboscis signifying an initially duplicated nasal placode.

Our patient exhibited an almost unique feature of an intraconal retrobulbar mass (1, 3, 7, 13-17). Intraoperatively, this was found to be a multilobulated cyst that contained clear fluid and arose from the optic nerve. This was not a cephalocele. The globe and lens were well developed but very small. Histologically, the cyst wall was composed of organized and disorganized retinal and glial tissue and the dura of the optic nerve enclosed the cyst. Thus, the possibility of a choristoma, a mass formed by maldevelopment of tissue of a type not normally found at that site, arose. The combined occurrence of microphthalmos with orbital cyst is a well-defined embryopathic entity felt to be secondary to incomplete

closure of the fetal embryonic (optic) fissure, which normally occurs in the sixth gestational week. There is a defect in the posterior globe in this complex. Typically, if an insult to the developing eye occurs before or shortly after the optic vesicle has developed from the forebrain in the fourth week, severe microphthalmos with a disordered globe, and subsequently a disordered lens, results (18, 19).

An intraorbital neuroectodermal cyst may occur with a well-ordered but small globe and without an obvious embryonic fissure defect. Although the ophthalmologic surgeon did not observe a connection between the globe and the cyst, the globe and cyst could not be operatively delivered intact, so that confirmation of fissure patency or closure could not be made pathologically. It is most likely that the orbital complex in our patient represented a "microphthalmia with cyst" which arose after 6 weeks and, therefore, may be temporally related to the defect creating the proboscis. Wang et al (3) previously reported a patient with proboscis lateralis who presented with an intraorbital cyst that they felt was secondary to "cystic degeneration of the optic nerve." The globe in their patient did not contain a lens and, therefore, is not the exact entity as in our case.

The complete intraoperative, histologic findings and surgical results are beyond the scope of this article. Briefly, the correction of this condition requires an incorporation of procedures geared to the correction of facial clefts, orbital wall, or cranial fossa defects. Therapy of the proboscis itself generally requires a two-stage procedure with the soft tissues of the proboscis acting as a pedicle flap for reconstruction of the defective nasal passage, with transection of the base and further cosmetic repair occurring at a separate setting (6, 7, 13).

Acknowledgments

We wish to express an appreciation to Maria Pembrook for her preparation of the manuscript and Martha Hefner for her medical illustration.

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