Pictorial Essay

Imaging Studies in the Diagnostic Workup of Neonatal Nasal Obstruction

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Abstract: Twelve neonates presenting with nasal obstruction after birth were evaluated by imaging studies for diagnostic reasons. Four groups were recognized: Group I: choanal atresia (n = 5) and choanal stenosis (n = 1); Group II: congenital nasal pyriform aperture stenosis (CNPAS) (n = 3) and holoprosencephaly (n = 1); Group III: nasolacrimal duct mucocele (n = 1); Group IV: nasal hypoplasia (n = 1). Associated anomalies were found in eight patients. Four patients with choanal atresia showed manifestations of the CHARGE (coloboma, congenital heart defect, atretic choanae, retarded physical and neuromotor development associated with central nervous system anomalies, genital hypoplasia, and ear anomaly and/or deafness) association. In the fifth patient with choanal atresia, the diagnosis of amnion disruption sequence was made. One patient with CNPAS had a solitary maxillary central incisor (SMCI), a mild form of holoprosencephaly. Besides proboscis and synophthalmos, SMCI was also present in the holoprosencephaly case. The patient with severe nasal hypoplasia had warfarin embryopathy. This review emphasizes the need for performing imaging studies in the diagnostic workup of neonates born with nasal obstruction.

Index Terms: Neonates—Nasal obstruction—Choanal atresia—Computed tomography—Magnetic resonance imaging.

Congenital choanal or nasal anomalies are uncommon but have the potential of life-threatening airway obstruction in the neonate. They may be the result of nasal cartilage and/or nasal bone hypoplasia, bony inlet or pyriform aperture narrowing, septal displacement, septal hematoma, nasal masses, and narrowing or atresia of the posterior choanae. Clinically, nasal obstruction can be diagnosed by failure or great difficulty in passing a 3- to 4-mm-thick nasogastric tube or by the presence of respiratory distress at birth and/or cyanosis during feeding. This article documents the radiologic findings in patients with neonatal nasal obstruction and also provides the opportunity of highlighting, with imaging procedures, the anomalies associated with the airway obstruction.

DIAGNOSTIC APPROACH AND PATIENT POPULATION

Specification of the anatomic site of the nasal airway obstruction using CT of the nasal cavity, the nasopharynx, and orbits can be obtained using contiguous 3-mm-thick slices in a plane parallel to the hard palate. Measurements should be performed at the level of the maximum transverse diameter of the pyriform aperture, the posterior choanae, and the posterior inferior os vomer. The pyriform aperture is considered stenotic when either the maximum transverse diameter of each aperture is \( \leq 3 \) mm (1) or when the pyriform aperture width is <8 mm (2). Choanal atresia can be diagnosed if the posterior choanal orifice measures \(<0.34\) cm unilaterally, in combination with a thickened inferior posterior os vomer measuring \( \geq 0.55\) cm (3). Whenever possible, we perform a CT examination of the temporal bone and an MR examination of the brain and inner ear [3D Fourier transformation–constructive interference in steady state (3DFT-CISS)-weighted images] in patients with choanal atresia. Skeletal X-ray examinations can be helpful in case of multiple malformations.
Nasal obstruction can result from several different anomalies. In Table 1, relevant clinical history and nosologic diagnosis, anatomic level of obstruction, additional imaging findings, and imaging procedures in 12 patients are summarized. By nasal cavity CT, the level of airway obstruction was demonstrated in all patients but Patient 10 with holoprosencephaly, proboscis, synophthalmal, and absence of nasal cavity. Based on the nasal and orbital CT findings, four anatomic types of neonatal nasal obstruction may be discerned: Group I: Patients 1–6, choanal atresia or stenosis of the choanal orifices; Group II: Patients 7–10, pyriform aperture stenosis/ holoprosencephaly; Group III: Patient 11, nasolacrimal duct mucocele; Group IV: Patient 12, nasal hypoplasia.

**DISCUSSION**

**Group I: Choanal Atresia/Choanal Orifice Stenosis**

Choanal atresia or choanal orifice stenosis (Figs. 1–5) is an airway obstruction caused by significant narrowing of the posterior nasal passage(s) or choana(e). In 1910, Fraser described choanal atresia in 115 cases, in 90% of whom the obstruction was bony and in 10% membranous. In a recent review of 63 CT examinations in patients with choanal atresia, fewer than one third were classified as pure bony and over two thirds were mixed bony-membranous, but no pure membranous forms were identified (4). These numbers better reflect the anatomic anomalies. The ratio of unilateral to bilateral choanal atresia has been estimated to be 2:1. The average incidence of the anomaly is 0.82/10,000 newborns (5).

Choanal atresia or stenosis may either be an isolated clinical and radiologic finding or be part of a syndrome of multiple congenital malformations (6–8). In fact, it has already been recognized as a more or less consistent component in >20 syndromes (7). Rare familial instances do occur (9). The anomaly has also been reported following in utero methimazole exposure (10,11). In 6% of choanal atresia patients, the karyotype has been abnormal. In 5%, a monogenic cause has been identified or suspected. Associated anomalies are encountered in 47% of patients, without detectable karyotype abnormality (6). Some of these patients represent examples of the CHARGE association of congenital anomalies. This acronym stands for coloboma (C), congenital heart defect (H), atretic choanae (A), retarded physical and neuromotor development associated with CNS anomalies (R), genital hypoplasia (G), and ear anomaly and/or deafness (E). Clinical recognition of the CHARGE association requires the presence of at least four of the congenital anomalies. In Table 1, relevant clinical history and nosologic diagnosis, anatomic level of obstruction, additional imaging findings, and imaging procedures in 12 patients are summarized. By nasal cavity CT, the level of airway obstruction was demonstrated in all patients but Patient 10 with holoprosencephaly, proboscis, synophthalmal, and absence of nasal cavity. Based on the nasal and orbital CT findings, four anatomic types of neonatal nasal obstruction may be discerned: Group I: Patients 1–6, choanal atresia or stenosis of the choanal orifices; Group II: Patients 7–10, pyriform aperture stenosis/ holoprosencephaly; Group III: Patient 11, nasolacrimal duct mucocele; Group IV: Patient 12, nasal hypoplasia.

**TABLE 1. Twelve patients with congenital nasal obstruction: summary of clinical and imaging data**

<table>
<thead>
<tr>
<th>Patient no./gender</th>
<th>Clinical history, nosologic diagnosis</th>
<th>Anatomic obstruction level</th>
<th>Additional imaging findings</th>
<th>Procedure</th>
</tr>
</thead>
<tbody>
<tr>
<td>Group I</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>1/M</td>
<td>Unknown</td>
<td>Choanal stenosis (borderline pathologic measurements)</td>
<td>Absent</td>
<td>CT</td>
</tr>
<tr>
<td>2/M</td>
<td>Antiepileptic drugs during pregnancy, exitus due to left heart hypoplasia, CHARGE</td>
<td>Unilateral left, bony-membranous choanal atresia</td>
<td>Absent semicircular canals, bilateral coloboma</td>
<td>CT(+), MR(+)</td>
</tr>
<tr>
<td>3/F</td>
<td>CHARGE</td>
<td>Bilateral, bony-membranous choanal atresia</td>
<td>Absent semicircular canals, absent corpus callosum</td>
<td>X-ray, CT(+), MR(+)</td>
</tr>
<tr>
<td>4/F</td>
<td>CHARGE</td>
<td>Unilateral, bony choanal atresia</td>
<td>Absent semicircular canals, bilateral coloboma</td>
<td>X-ray, CT(+)</td>
</tr>
<tr>
<td>5/M</td>
<td>CHARGE</td>
<td>Unilateral left, bony-membranous choanal atresia</td>
<td>Absent semicircular canals, right-sided coloboma, absent 12th ribs, four sacral segments</td>
<td>X-ray, CT(+)</td>
</tr>
<tr>
<td>6/M</td>
<td>Amnion disruption sequence</td>
<td>Bilateral, bony choanal atresia</td>
<td>Chiari I, left-sided coloboma, amputation of fingers and toes</td>
<td>X-ray, CT(+), MR(+)</td>
</tr>
<tr>
<td>Group II</td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>7/F</td>
<td>CNPAS</td>
<td>Pyriform aperture stenosis (5.9 mm)</td>
<td>Absent</td>
<td>CT</td>
</tr>
<tr>
<td>8/F</td>
<td>Homeopathic sedatives during pregnancy, CNPAS</td>
<td>Pyriform aperture stenosis (5.2 mm)</td>
<td>Absent</td>
<td>CT</td>
</tr>
<tr>
<td>9/F</td>
<td>Intrauterine deeded co-twin, hydropydrum, 33W, hydroureteronephrosis, CNPAS</td>
<td>Pyriform aperture stenosis (4.9 mm)</td>
<td>SMCI</td>
<td>X-ray, CT</td>
</tr>
<tr>
<td>10/F</td>
<td>Holoprosencephaly</td>
<td>Absent nasal cavity</td>
<td>SMCI</td>
<td>X-ray, MR(+)</td>
</tr>
<tr>
<td>Group III</td>
<td></td>
<td></td>
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<td></td>
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<tr>
<td>11/F</td>
<td>Dacryocystitis (left), nasolacrimal duct mucocele</td>
<td>Cyst inferior to left lower turbinate</td>
<td>Absent</td>
<td>CT</td>
</tr>
<tr>
<td>Group IV</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>12/F</td>
<td>Warfarin exposure, polyhydramnion, retarded development, conductive hearing loss</td>
<td>External nose skeleton, pyriform aperture (6 mm)</td>
<td>Chondrodysplasia punctata, brachydactyly, clubfoot</td>
<td>X-ray, CT, MR</td>
</tr>
</tbody>
</table>

CT, CT examination of nasal cavity and orbits; CT(+), CT examination of nasal cavity, orbits, and temporal bone; MR, MR examination of brain; MR(+), MR examination of brain and inner ear; CNPAS, congenital nasal pyriform aperture stenosis; SMCI, solitary maxillary central incisor; CHARGE, coloboma (C), congenital heart defect (H), atretic choanae (A), retarded physical and neuromotor development associated with CNS anomalies (R), genital hypoplasia (G), and ear anomaly and/or deafness (E).
anomalies (12). In a large study of choanal atresia patients, 7% of all patients and 17% of patients with multiple congenital malformations were considered examples with the CHARGE association. However, in the latter study, only three defects were required for the clinical diagnosis (6).

Ammnion disruption sequence is one of the several entities known to be associated with choanal atresia (7). Other component anomalies include transverse limb amputations and craniofacial clefts resulting from constricting amniotic band-like structures.

Therapy of choanal atresia consists of transnasal endoscopic resection or transpalatine surgical resection of the os vomer and stenting in the bony variants as restenosis may occur. Membranous choanal atresia may be treated by endoscopic perforation, followed by stenting (17).

Group II: Congenital Nasal Pyriform Aperture Stenosis/Solitary Maxillary Central Mega-Incisor/Holoprosencephaly

Congenital nasal pyriform aperture stenosis (CNPAS) (Figs. 6–9) was first described by Brown et al. (18) as a type of nasal obstruction in the newborn, caused by bony overgrowth of the nasal medial processus of the maxilla with the effect of narrowing of the bony inlet of the nasal cavity. While initially considered as an isolated abnormality, subsequently CNPAS was thought to represent a microform of holoprosencephaly, the result of abnormal development of the prosencephalon and midline facial structures. This hypothesis is based on the observation that a solitary maxillary central incisor (SMCI), a manifestation of holoprosencephaly, is found in many instances of CNPAS (19–21). Additional support for this hypothesis is provided by two twin pregnancies. In both, one of the children presented with holoprosencephaly while the other part of the monozygotic twin had CNPAS (22,23). SMCI was a feature in our tenth patient, with alobar holoprosencephaly, and in our ninth patient, the third child with CNPAS in the present series. Interestingly, our ninth patient is the surviving member of monozygotic twins, the co-twin having died in utero. Due to autolytic changes in the latter’s brain, holoprosencephaly could not be confirmed, but a single umbilical artery and esophageal atresia were found by postmortem examination. Holoprosencephaly is genetically heterogeneous as it is observed in various forms of aneuploidy but has, in addition, several monogenic or teratogenic causes (maternal diabetes). Clinically, it presents as a clinical continuum of phenotypes ranging from a mild hypotelorism, CNPAS, and SMCI to cyclopia and agenesis of the premaxilla in the more severely affected cases (20). The tenth patient is an example of the latter type of expression. Although SMCI and/or CNPAS may be isolated findings, pituitary hypoplasia and CNS abnormalities need ruling out in all patients (24).
Radiologic examination can demonstrate the anatomic site of airway obstruction in patients with CNPAS even in view of absent data on the normal dimensions of the pyriform aperture. CNPAS was diagnosed reliably by demonstrating bony overgrowth of the nasal processus of the maxilla on the axial or coronal CT images (5).

Therapy is conservative, using vasoconstrictive drugs and/or nasal stenting in the first weeks (25,26). If conservative therapy fails and the obstructive apnea leads to pulmonary hypertension and failure to thrive, surgical resection of the bony overgrowth at the level of the anteromedial maxilla can resolve the nasal airway stenosis.

FIG. 2. Patient 3. CHARGE. A: Axial CT image at the level of the posterior choanae shows bilateral bony-membranous choanal atresia (arrows). A thickened os vomer is present (arrowheads). B: Axial 1 mm CT sections through the left epi- and mesotympanum show a deformed vestibule (black arrowhead) and fixation of the malleus and incus (white arrows) to the anterior and lateral tympanic wall, respectively. A normal labyrinthine segment of the facial nerve (white arrowhead) is seen. No semicircular canals are visible. C: MR image resulting from maximum intensity projections of 1 mm slices performed with 3DFT-CISS sequence is shown. Semicircular canals are absent. The cochlea (arrows) and vestibule (arrowheads) are visible on both sides. D: MR performed with 3 mm sagittal T1-weighted sequence shows absence of the corpus callosum (arrows).

Group III: Lacrimal Sac Mucocele/Nasolacrimal Duct Mucocele

Congenital obstruction of the nasolacrimal duct is seen in up to 84% of newborns. The obstructed nasolacrimal system is filled with amniotic fluid and thus referred to as an amniocele. In 2–4% of the cases, the obstruction becomes symptomatic with bilateral involvement in 14% of the symptomatic cases. In 80% of these symptomatic patients, spontaneous rupture usually occurs within 6 months (27). The obstruction may, however, persist and give rise to accumulation of secretions in the amniocele with subsequent mucocele formation. For unknown reasons, the condition is seen more in the female, the
female/male ratio being ≈5:1 (28). Proximal failure of canalization of the nasolacrimal duct, at the level of the Rosenmuller valve, produces a distension of the lacrimal sac, leading to the condition of lacrimal sac mucocele, whereas distal failure of canalization at the level of the Hasner membrane produces a nasolacrimal duct mucocele (29).

Clinically, a bluish medial canthal mass and an intranasal submucosal cystic mass inferior to the lower turbinate may be present (30). Such bilateral intranasal masses may be responsible for respiratory distress in the newborn. In case of unilateral localization, the nasal

FIG. 3. Patient 4. CHARGE. A: Axial 1 mm CT section through the right mesotympanum shows a stump of the posterior semicircular canal or part of the vestibular aqueduct (arrowhead). The superior part of the vestibule (arrow) is also visible. B: Axial CT image through the orbits demonstrates bilateral posterior globe deformity (arrows), representing coloboma. C: The 3D surface reconstruction of the head shows the external ear deformity typical in patients with CHARGE association.

FIG. 4. Patient 5. CHARGE. A: Axial 1 mm CT section through the right mesotympanum shows fixation of the head of the malleus to the anterior tympanic wall (large arrow). A malformed incus is seen (small arrow). Widening and abnormal course of the facial canal (arrowheads) are present. B: Axial 1 mm CT section through the right mesotympanum shows opacification of the middle ear cavity. The stapes is absent. Note also the bony obliteration of the oval window (arrowhead). The tympanic sinus and pyramidal eminence are absent. The intercalar septum between the middle and apical turns of the cochlea is probably also absent. The state of the cochlear modiolus is doubtful (arrow).
FIG. 5. Patient 6. Choanal atresia-amnion disruption sequence. A: CT image performed at the age of 5 years at the level of the posterior choanae shows bilateral bony choanal atresia (arrows). B: MR image resulting from maximum intensity projection of 1 mm slices performed with 3DF-CISS sequence is shown. Normal posterior and lateral semicircular canals are visualized (arrows). C: Sagittal T1-weighted MR image performed at the age of 5 years shows the obstruction of the choanae (arrowhead) and Chiari I malformation (arrow). The Chiari I malformation was not present at birth. D: Partial amputation of the fingers with convergence and tapering of the phalanges (arrows) supports the diagnosis of amnion disruption sequence. E: MR image performed with 4 mm coronal T2-weighted sequence shows an outpouching at the level of the posterior-inferior side of the globe, representing a coloboma on the left side (arrow).
cycle may obstruct the normal side when vasocongestion of the nasal mucosa occurs (27). Infection may lead to pyocele formation (31). In the literature, no associated anomalies have been described.

Both CT and MR are effective in demonstrating the characteristic triad of an enlarged lacrimal fossa due to the distension of the lacrimal sac, a dilated nasolacrimal duct, and a cystic mass inferior to the lower turbinate (Fig. 10). Moreover, these imaging techniques allow the differentiation of this condition from other forms of nasal

airway masses such as nasal glioma, meningoencephalocele, and other masses within the nasal cavity. In the orbits, dacryocystitis, lymphangiohemangioma, and dermoid need ruling out (30,32).

Nasolacrimal duct probing may suffice as treatment of proximal distensions without cystic nasal mass. In case of nasal protrusion, endoscopic marsupialization is necessary (27,28,33).

Group IV: Nasal Hypoplasia

Chondrodysplasia punctata (CP) (Fig. 11) is the term used for a heterogeneous group of skeletal dysplasias characterized by epiphyseal extraosseous stippling. These epiphyseal abnormalities tend to disappear in the first 3–5 years of life, rendering diagnosis harder later on in life. CP was first described by Conradi in 1914. The term Conradi-Hünermann disease is frequently but indiscriminately used as a label for this etiologically heterogeneous group of diseases (34). Causes of this form of skeletal dysplasia include various metabolic, karyotypic, and teratogenic disorders including warfarin embryopathy (35). In vitro studies have shown that high concentrations of warfarin inhibit the catalytic activity of arylsulfatase E. The gene encoding this enzyme protein harbors mutations in patients with the brachytelephalangic type of CP (36,37). Therefore, the phenotypes of warfarin embryopathy and of the brachytelephalangic type of CP are almost congruent, as both show frontal bossing, flat nasal bridge, optic nerve atrophy, cataract, short neck, short limbs, brachydactyly, low birth weight, and
respiratory difficulties (7,38). Owing to the underdeveloped cartilaginous skeleton of the nose, the anterior part of the nasal septum and the nasal wings are usually hypoplastic.

Unsupported by cartilage, the soft tissues may obliterate the pyriform aperture, with subsequent respiratory problems. Shortening of the columella and depression of the nasal bridge compound the potential airway obstruction, possibly worsened still either by co-existent choanal stenosis, stenosis of the trachea and bronchi, or a stenotic pyriform aperture.

Conventional radiographs demonstrate epiphyseal calcifications in the first years of life. CT may be helpful in determining the exact site of airway obstruction within the nasal cavity and elsewhere such as the trachea. Because of the associated conductive hearing loss we observed, temporal bone imaging may be performed to confirm or exclude anomalies of the middle or inner ear structures.

Although nasal airway obstruction usually resolves in older children owing to nasal airway expansion through growth, there is also growth-dependent improvement of the nasal bridge depression. However, initial stenting of the nasal airway is often required. Tracheal cannulation may be needed (39).

CONCLUSION

In conclusion, the finding of neonatal nasal obstruction requires additional imaging studies to rule out associated anomalies. The latter were observed in two thirds of the group of 12 patients presented. Also, in patients with choanal atresia, high resolution CT scan of the temporal bones may be helpful to rule out the CHARGE association of congenital anomalies by the demonstration of aplasia of the semicircular canals.

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REFERENCES


**FIG. 10.** Patient 11. Nasolacrimal duct mucocele. A: A left-sided medial canthal swelling is present in this patient with nasolacrimal duct mucocele. B: CT performed with 2 mm axial sections at the level of the orbits shows enlarged lacrimal fossa (arrows). C: Reconstructed coronal CT image shows an enlarged nasolacrimal duct (arrowheads). D: CT performed with 2 mm axial sections inferior to the lower nasal conchae shows a unilateral cyst on the left side (arrow).


**FIG. 11.** *Patient 12. Nasal hypoplasia. A:* Stippled epiphyses are seen at the level of the shoulders and around vertebral bodies in this patient with chondrodysplasia punctata secondary to intrauterine warfarin exposure. *B:* The 3D surface reconstruction of the face demonstrates severe nasal hypoplasia. *C:* Axial CT image at the level of the pyriform aperture shows nasal hypoplasia with combined soft tissue-bony obstruction at the level of the pyriform aperture (arrows).