Airway management in Nager Syndrome

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Abstract

Nager acrofacial dysostosis is a rare congenital syndrome characterized by malformations of the mandibulofacial structures and pre-axial upper limbs. Patients classically present with malar hypoplasia, micro/retrognathia, anti-mongoloid slanting, cleft deformities, ear anomalies, and hypoplastic thumbs. Trismus and glossoptosis from mandibular abnormalities predispose infants to life-threatening respiratory distress after birth. A case of a Nager Syndrome mother delivering a similarly afflicted fetus is presented, with approaches to maintaining both tenuous airways described. Nager Syndrome pts warrant meticulous airway management upon delivery.

Introduction

ALR is a 32 year old, G1P0 female with Nager Syndrome. A fetal ultrasound identified findings consistent with Nager Syndrome (Fig 1a). ALR had findings of dysplastic pinnae, retro/micrognathia, downward slanted palpebral fissures, maxillary hypoplasia, cleft palate, phocomelia, and kyphoscoliosis. The patient had undergone multiple maxillofacial surgeries as a child, including cleft palate repair, chin implant, bone-anchored hearing aid implantation, as well as a total spinal fusion with placement of Harrington rods. She had a known history of difficult intubations. There was no other known family history of Nager Syndrome, mental retardation, consanguinity, or birth defects.

ALR presented at 34 5/7 weeks gestation with preterm premature rupture of membranes. Due to her difficult airway, regional anesthesia was first attempted but failed, and ultimately fiberoptic intubation was successfully performed for airway establishment.

Clinical course

AJR was delivered via Cesarian section and was found to be apneic and cyanotic, with APGARs of 2 (1 minute), 2 (5 minutes), and 5 (10 minutes). Birth weight was 1.9kg (25th percentile) and length was 46cm (50th percentile). Three attempts at orotracheal intubation were unsuccessful, with the patient became progressively bradycardic and hypoxic by skin color despite mask ventilation. An emergency tracheostomy was then performed successfully at 15 minutes of life (Fig 1b).

Table 1: Conditions similar to Nager Syndrome.

<table>
<thead>
<tr>
<th>Comparable Syndrome</th>
<th>Differentiating Factors</th>
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<tbody>
<tr>
<td>Edwards Syndrome (Trisomy 18)</td>
<td>karyotype, clinodactyly</td>
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<tr>
<td>Treacher Collins Syndrome</td>
<td>no limb/digit anomalies; MR; commonly AD</td>
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<tr>
<td>Mohr Syndrome (oral-facial-digital syndrome)</td>
<td>polydactyly or bifid allux</td>
</tr>
<tr>
<td>Roberts Syndrome</td>
<td>karyotype (premature centromere separation, phocomelia)</td>
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</tbody>
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AJR was found to have low-set dysplastic pinnae, stenotic external auditory canals, downslooting palpebral fissures, malar hypoplasia, retro/micrognathia, high-arched cleft palate, shortened forearms, bilateral clinodactyly, bilateral club feet, an absent/hypoplastic thumbs. Skeletal survey (Fig. 2) found proximal radial-ulnar synostosis.

AJR subsequently underwent a bilateral mandibular osteotomy with distraction at 3 months of age, and a gastrostomy tube placement at 4 months of age.

Conclusions

While not all Nager Syndrome patients will require tracheostomy, the more severe degree of mandibular and palatal defects suggest that prenatal diagnosis is vital. The vulnerable airway mandates close monitoring by specialty services equipped to secure it. Airway options include prone positioning, nasopharyngeal airway, intubation, and tracheostomy. Once the infant is stabilized, a multidisciplinary team is best suited to care for this challenging disorder.