Clinical Considerations in a Patient with Nager Syndrome & Pierre Robin Sequence: A Case Report

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NAGER SYNDROME

Nager syndrome is a rare genetic disorder involving mutations of the *SF3B4* gene. Incidents are generally considered sporadic, however, there have been some reports of an autosomal dominant or recessive inheritance. The condition can present with a variety of abnormal clinical manifestations including the following:

Upper extremities: radial hypoplasia or aplasia, ulnar dysplasia, thumb hypoplasia or aplasia, and a "bowed" arm appearance

Craniofacial: downward slanting eyes, micrognathia, and aplasia of the mandibular ramus and TMJ

Otolaryngeal: atresia of external ear canal and auricle causing conductive deafness, hypoplasia of the larynx and epiglottis

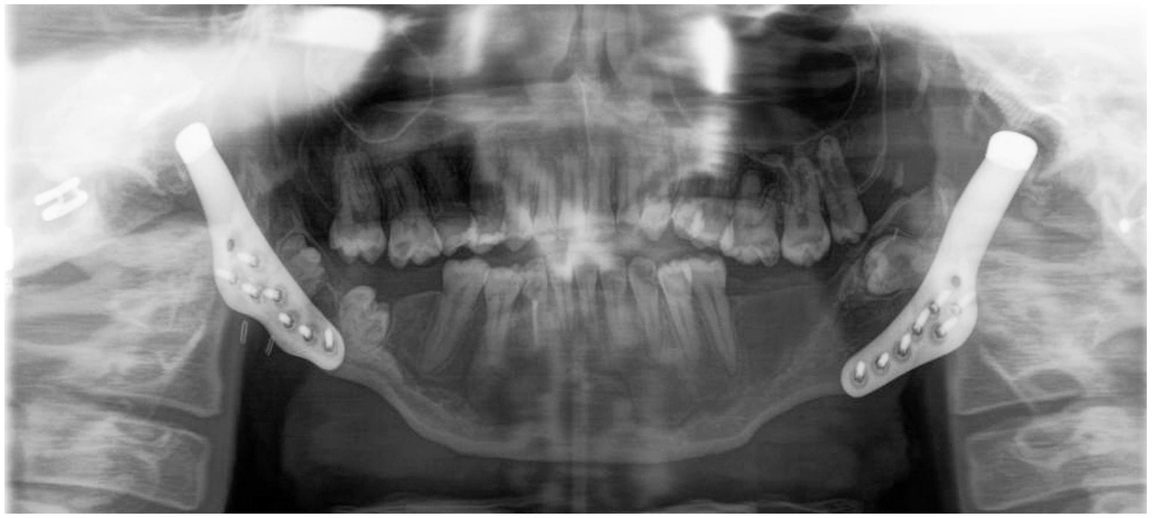
Systemic: tetralogy of Fallot, unilateral renal agenesis

Many patients with Nager syndrome require tracheostomy at a young age due to facial anomalies causing airway obstruction. Feeding requires supplementation either through a G-tube or liquified diet due to limitations in jaw movement and function. Speech delays are often noted as a consequence of hearing loss; however, the syndrome itself does not have any negative impact on intelligence. Those who survive past infancy retain an otherwise normal growth pattern leading to an overall healthy stature.[1]

PIERRE ROBIN SEQUENCE

Pierre Robin Sequence is a congenital malformation characterized by the presence of 3 craniofacial anomalies: (1) cleft palate, (2) mandibular micrognathia, and (3) glossoptosis. Mandibular micrognathia and retrusion contributes to the patient's glossoptosis by causing posterior displacement of the tongue with a reduction in sublingual musculature that supports tongue protrusion, ultimately leading to airway obstruction. Cleft palate results in feeding difficulties associated with abnormal suckling patterns and malocclusions from a collapsed maxillary arch.[2] Airway obstructions are initially addressed through prone positioning and/or nasal airways with positive airway pressure. Surgical treatment can include tongue-lip adhesion to tie the tongue in a more anterior position, and distraction osteogenesis or orthognathic surgery to correct the micrognathic mandible.[3]

PANORAMIC RADIOGRAPH



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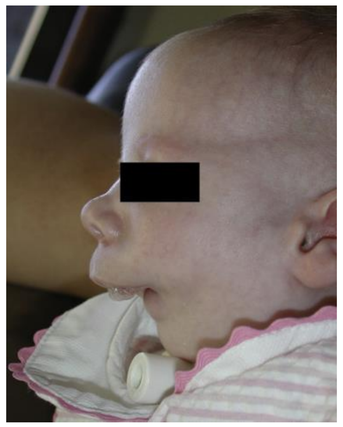
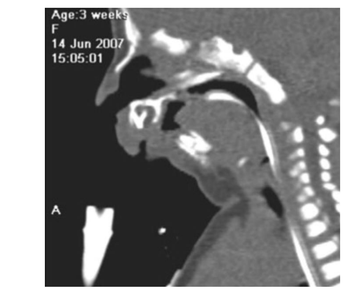
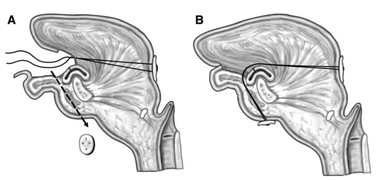
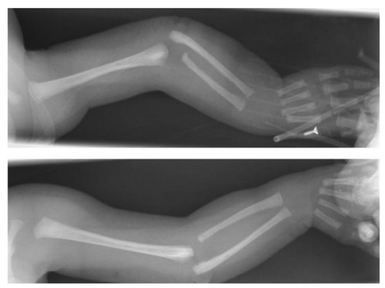


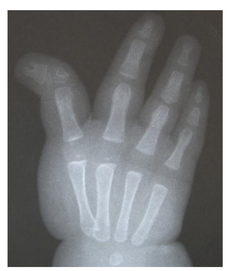
A patient with Nager syndrome demonstrating mandibular micrognathia and retrusion.[1]





Top: CT of a patient with glossoptosis resulting in airway obstruction.[4]
Bottom: Tongue-lip adhesion procedure.[5]





Top: Bowing of limbs and ulnar dysplasia.[1]
Left: Thumb aplasia.[1]

CLINICAL PRESENTATION

A 20-year-old female patient presented to the Craniofacial Clinic at Riley Children's Hospital Outpatient Center for evaluation with a chief complaint of: "she has very limited mouth opening and needs her joints replaced". A comprehensive history was obtained with a record of previous surgeries including tracheostomy and gastrostomy tube placement, bilateral temporomandibular joint prostheses, cleft lip and palate repair, thumb reconstruction, and bone-mounted hearing aid devices all performed at other institutions. The patient's range of motion was considered adequate immediately following the bilateral TMJ prostheses placement approximately 10 years ago. However, recurrent infections and progressive trismus have continued to limit the patient's mandibular range of motion to approximately 3mm of opening. Due to the inability to perform basic oral hygiene practices, there was significant plaque and calculus accumulation throughout the dentition. Intraoral radiographs could not be obtained. A panoramic radiograph presents the bilateral TMJ prostheses, caries on multiple teeth, and unerupted mandibular posterior teeth. Despite significant challenges in daily living, the patient is pursuing higher education and is a part of a sorority.

TREATMENT/MANAGEMENT

The patient has been seen by multiple specialists at Indiana University School of Dentistry including pediatric dentistry, endodontics, and oral and maxillofacial surgery (OMFS). A pulpectomy was attempted on tooth #12 but ultimately could not be completed due to limited access, and a sedative restoration was placed. The patient was evaluated by multiple specialists of the Craniofacial Clinic who are working together to address the needs of the patient. Pediatric dentistry has provided routine maintenance care as best as possible considering the patient's limited opening. A comprehensive treatment plan is still being considered by the multidisciplinary team with OMFS taking the lead role. However, it has been discussed that modification/replacement of the TMJ prostheses is unlikely to result in a more favorable outcome.

REFERENCES

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