

Severe Agnathia–otocephaly complex : Surgical Management & Longitudinal Follow-up of 4 patients from birth through adulthood.

Michael Alperovich MD, Michael S. Golinko MD, MA, Pradip Shetye DDS, BDS, MDS, Roberto L. Flores MD, David A. Staffenberg MD DSc(Hon)

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Introduction: Agnathia–otocephaly complex (AOC) is characterized by mandibular hypo- or aplasia, ear abnormalities, microstomia, and microglossia. The spectrum of this rare syndrome extends from patients born without a mandible, agnathia-- to patients with micrognathia. In between these two, we observed a *nanognathia*: absence of body *and* ramus, and only a midline symphyseal remnant. The literature reports on only seven patients with severe AOC whom have survived past infancy.¹⁻⁵ We report our single-institution, long-term follow-up on 4 patients, present our reconstructive experience and suggest a management strategy.

Methods: A single institution review of all patients with AOC treated over a 30 year period was reviewed.

Results: Four patients were identified, one female had agnathia, two males had *nanognathia* and the remaining male had micrognathia. All were followed at birth or prior to one year of age. The mean follow-up was 17 years (6- 28). Peri-natal findings included polyhydraminos on pre-natal ultra-sound, premature birth and perinatal tracheostomy and gastrostomy-tube placement. Soft tissue findings included severe microstomia, choanal atresia and absence of tongue. Commissuroplasties were typically performed before 3 years of age and repeated as necessary to allow for oral hygiene and access for surgery. De novo genioplasty was performed with rib between ages 3 and 8, after which time, free fibula transfer was utilized. Due to resorption or extrusion, all patients underwent repeat bone grafting. Tissue expansion of the neck was commonly used to augment soft tissue of the lower face, but was more successful in the teenage years. Although all patients retained tracheostomy and gastrostomy-tube dependent, they were able to communicate, make friends and attend school.

Conclusions: AOC need not be a fatal nor untreatable condition; a reasonable quality of life can be achieved. Although the lower-facial contour may be restored, and stoma created, the inherent lack of musculature make deglutition virtually impossible with current therapies. Just as transplantation has emerged as a modality for facial restoration following severe trauma, so too may it be a future option for congenital deformities.

References

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