Critical Phenotypic Manifestations of 22q11.2 Deletion Syndrome In Patients Requiring Surgery for Velopharyngeal Insufficiency

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Abstract Text:

Purpose
Patients with 22q11.2 deletion syndrome (22q) exhibit an extremely variable phenotype. In particular, cervical vascular abnormalities are important to recognize preoperatively in patients undergoing procedures for velopharyngeal insufficiency (VPI). We sought to identify the frequency of specific cardiac abnormalities, cervical spine instability, and aberrant neck vasculature in our population of patients with 22q undergoing surgery for VPI, and aimed to evaluate whether specific deletions at the 22q locus were associated with the radiographically recognized phenotypic manifestations.

Methods
A retrospective chart review of all patients with 22q who underwent VPI surgery was performed. Those who had preoperative cervical MRA or CTA, cervical spine imaging, and cardiac evaluation were included in this study. In addition, the genetics department provided the specific genetic deletion for 38 patients in our study.

Results
81 patients with 22q undergoing VPI surgery were recorded. 70 patients had imaging available for review. 39 (55.7%) patients had at least one vascular abnormality of the neck, 24 patients (34.3%) had carotid abnormalities, most commonly a low bifurcation(11) or medial deviation of the internal carotid(11). 28 patients(40%) had other vascular abnormalities, most commonly an aberrant course of the neck vessels(12) or an abnormal arch origin(8). In one patient the carotid and vertebral anatomy was so complex that surgery was not performed. Cervical spine abnormalities were found in 22(31.4%) patients. 16(72.7%) of these had vascular abnormalities of the neck and 10(45.5%) had carotid abnormalities. Carotid abnormalities occurred most commonly with hypoplastic C1(50%) and fusion of one or more cervical levels(57%). Cardiac abnormalities were found in 46(65.7%) patients. 28(60.9%) of these patients had abnormal neck vasculature, and 17(37%) had carotid abnormalities. Carotid abnormalities were most common in the presence of a VSD(46.2%) and a right aortic arch(44%) and least common in TOF(21.4%).

38 patients had evaluation of their genetic deletion. 34(89.5%) possessed the typical deletion(A-D). 2(5.3%) had the atypical nested(A-B) deletion, and 1(2.6%) had(B-D) deletion. One patient had a 1.4mb deletion. There was no significant difference in findings between different deletion size groups.

Conclusion
Preoperative diagnosis of cervical spine, vascular, and cardiac pathology is essential to prevent avoidable surgical complications in patients with 22q undergoing VPI surgery. Deletion size does not correlate with phenotypic findings in this study, but further studies are necessary to solidify that conclusion.