Congenital Cataract Surgery and SOX2 Gene Evaluation in Microphthalmic Eyes

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Introduction. In children congenital cataracts represent 25% to 30% of avoidable blindness, and microphthalmus has been found in 7% to 17% of these patients. Our purpose is to report the outcomes and complications of congenital cataract sugery with Intraocular-Lens(IOL) implantation in microphthalmic eyes. In microphthalmic eyes IOL implantation is more challenging due to small anatomy, but it gives better visual prognosis than eyes left aphasic. SOX2 gene has been identified as a major causative gene of microphthalmus. Screening of SOX2 was performed.

Aim, Material and Methods. The study involved 20 microphthalmic eyes from 14 children younger than 3 years of age with congenital cataract. Surgeries consisted in aspiration of the lens and IOL implantation. Eyes with inflammation, ocular trauma, aniridia, chorioretinal coloboma or vitreoretinal diseases were excluded. 6 patients had bilateral cataract. The outcome measures were Intraocular Pressure (IOP), Best-Corrected Visual Acuity (BCVA) intraoperative and postoperative complications. SOX2 coding region was amplified and PCR product were sequenced with Big Dye Terminator v3.1.

Results. Mutations in SOX2 account for 10–20% of A/M. Individuals with SOX2 mutations often have associated systemic anomalies, termed SOX2 anophthalmia syndrome, consisting of ocular, brain, pituitary, genitourinary, and gastresophageal anomalies, although eye defects can be isolated as well. In our study, mean age at the time of surgery was 23.9 ± 1.7 months. Mean ocular axial length was 18.6 ± 0.7 mm. Mean preoperative IOP was 9.3 ± 1.2 mmHg and 10.8 ± 2.7 mmHg on final follow-up. No one intraoperative complications happened. The post-operative complications observed were irregularity of pupil in 2 eyes, glaucoma in 3 eyes, posterior synechiae in 2 eyes, visual axis obscuration due to posterior capsule opacification (PCO) in 1 eye and phthisis in 1 eye (the shortest one with 17.9 mm of axial length). Pre-operative and post-operative BCVA was 2.01 ± 0.89 logMAR and 0.31 ± 0.06 logMAR in bilateral cases and 1.81 ± 0.97 logMAR and 0.32 ± 0.13 logMAR in unilateral cases, respectively. SOX2 mutations appear in 3/14 cases (15%). Most of the SOX2 coding region mutations are represented by nucleotide substitutions or deletions/insertions resulting in a premature truncation of the normal protein. The majority of these truncating mutations are observed de novo in patients. Several missense mutations in SOX2 have also been identified and are predicted to affect the DNA-binding or transactivation domains of this protein, which are critical to its activity. Long term improvement of vision is under control and children will be followed-up every 3 to 6 months.

Conclusions. In microphthalmic eyes primary IOL implantation in congenital cataract surgery resulted in a significant BCVA improvement with minimal postoperative and no intraoperative complications. Glaucoma and axial opacifications are well-known long term postoperative complications and children who underwent congenital cataract surgery must be followed-up regularly. Shorter axial lenght is a significat risk factor for post-operative complicatons. SOX2 mutation is found to be one of the major causes of microphthalmus and further studies of mutations affecting the lens membranes (aquaporins/Mip, Lim-2 or connexins) or the structural proteins of the cytosol will give a better uderstanding of congenital cataracts in developing eyes.