Brief Communication

Paediatric Syndromes with Craniofacial Abnormalities and Ophthalmic Manifestations – Anaesthetic implications

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Paediatric syndromic patients with eye disorders frequently require anaesthetic management for repeated ophthalmic examinations and surgical treatments or both. These patients present a challenge for the anaesthesiologists particularly regarding airway management or systemic stability throughout the procedure.[1] These syndromes with ophthalmic presentations may be categorized in five groups as phacomatoses, connective tissue disorders, chromosomal anomalies, metabolic storage diseases and craniofacial abnormalities. Each group has specific manifestations that may affect procedural anaesthesia practice.

In this issue of the journal, Catalca and et al. highlighted Saethre-Chotzen syndrome with craniofacial abnormalities focusing on its possible challenges for the anaesthesiologists.[2] In these patients, the airway management and vascular access might be predominantly difficult.

Other than Saethre-Chotzen syndrome, there are various conditions under the classification of craniofacial abnormalities. Hypertelorism, telecanthus, craniosynostosis, the first and second brachial arch defects are frequently associated conditions with ocular manifestations which are also very familiar conditions for the anaesthesiologists in name and management. While ***Apert Syndrome, Pfeiffer Syndrome and Crouzon Disease*** are the most common examples of disorders with craniosynostosis, Goldenhar Syndrome and Treacher Collins Syndrome are mandibulofacial dysostoses related to the first and second brachial arch defects. There are several rare disorders with ocular findings under this group.

Apert Syndrome is caused by the synostosis of coronal suture with the phenotypic features such as mid-facial hypoplasia, high arched or cleft palate, lower jaw protrusion

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which all may, in turn, complicate safe airway management. These patents have shallow orbits, severe proptosis, globe subluxation, strabismus as esotropia with a V-pattern. They usually present for strabismus surgery and reconstructive procedures. They may present with supraglottic airway obstruction by the rate of 4.5%.[3] However, infraglottic anomalies may also exist. An extensive plan for tracheal intubation should be available since supraglottic airway devices may not fit or work properly.[4] Intraoperative hypoglycaemia and hyponatremia, sometimes persistent, should always be considered in these patients.[5] Other than these precautions, due to abnormal orbit and globe manifestations, the non-operated eye should be conserved highly to avoid any eye injury during the surgery.

Very similar to Apert Syndrome, patients with ***Pfeiffer syndrome*** have extremely shallow orbits and severe proptosis with corneal exposure. They have characteristic broad and short thumbs and toes. Due to respiratory distress because of severe nasal obstruction, these patients may require neonatal tracheostomy and may present to operation room with their permanent canula which should be replaced during the anaesthetic management with an endotracheal tube or another tracheostomy canula which is compatible with the surgery.[6] Laryngeal web may observed.[7] They frequently have obstructive sleep apnea and related postoperative risks. They commonly present for strabismus surgery and the same preparations regarding airway management with more sophisticated equipment such as fiberoptic gadgets and non-operated eye preservation should be in place while these patients undergo an eye operation.[8]

Crouzon Disease, also similar to the previously mentioned syndromes, manifests with severe proptosis, corneal opacity due to exposure and optic nerve compression, iris coloboma, microcornea, ectopia of lentis, maxillary hypoplasia, hook-shaped nose, flattened forehead, respectively large tongue and deafness. Brainstem herniation and Chiari I syndrome may be observed due to small cranial vault. Glaucoma and strabismus are very common in these patients. Securing the airway is expected to be difficult and requires a preparation according to difficult airway algorithm guideline. Obstructive sleep apnea risk should be evaluated preoperatively. Sedatives and opioids should be avoided while postoperative chest physiotherapy and CPAP should be employed.[9] Possible increase in intracranial pressure due to abnormal head shape should not be aggravated by hypoventilation and hypercarbia.[10]

Goldenhar Syndrome (Oculoauriculovertebral-spectrum) shows typical features such as hemifacial microsomia and asymmetry, mandibular hypoplasia, cleft lip and palate, external, middle and internal ear malformations and

hearing disorders, vertebral anomalies such as scoliosis, spina bifida and fusion defects. Brainstem compression, cerebrospinal fluid disruption can be present. Eye involvement include limbal dermoids, epibulbar or conjunctival lipodermoids, lid dermoids. These features are bilateral in Goldenhar Syndrome and may be along with malformations of heart, lung, and kidney.[11] The upper airway manifestations of Goldenhar syndrome range from malocclusion to temporomandibular joint ankylosis making mouth aperture limited.[12] These patients may have difficult airway management, atropine-resistant bradycardia and impaired renal function.

Treacher Collins Syndrome (Mandibulofacial dysostosis) has bilateral ophthalmic manifestations such as eyelid coloboma, absence of lacrimal punctum, lower lid notch, ptosis along with systemic signs that may complicate airway management. These abnormalities include hypoplasia of malar and mandibular bones, cleft palate, blind fistula between mouth and ears. Their upper airway obstruction may worsen by age and direct laryngoscopy becomes more difficult concomitantly.[13] However, there are papers in the literature depicting successful use of supraglottic airway equipments. The level of anomalies at presentation for preoperative evaluation ensures anticipating possible perioperative complications. Therefore, further radiologic imaging and multidisciplinary team involvement should be planned before the surgery in difficult cases.[14]

The other rare disorders with ocular findings include, but not limited to, Aicardi, Cockayne, Rubinstein-Taybi, Hallermann-Streiff and Fetal Alcohol Syndromes.

Aicardi Syndrome presents with strabismus, nystagmus, microphthalmos, optic nerve hypoplasia and ptosis, and skeletal anomalies such as spina bifida, cleft lip and palate, fused ribs and vertebral anomalies. However, it is predominantly significant due to the progressive neurological deterioration such as mental retardation, seizures and hypotonia and death at childhood.[15] Neuromuscular agent effect is to be prolonged. Seizure prevention is recommended. These patients may require anaesthetic management for supportive measures since there is no specific treatment.

Patients with Cockayne Syndrome typically have premature aging and dwarfism. Facial features include bird like appearance and eye disorders such as nystagmus, corneal opacification, cataracts, band keratopathy, poor pupillary response. While neurological sequelae such as mental retardation, cerebellar ataxia and neurosensorial deafness require general anaesthesia or sedation in these patients for every intervention, muscle rigidity and seizures complicate the anaesthesia management.[16] Unfortunately, mortality is expected during late adolescence in Cockayne Syndrome. These patients may have myocardial ischemia and delayed recovery after anesthesia.[17]

Rubinstein-Taybi Syndrome features short stature, mental retardation, broad thumbs and toesantimongoloid slant of palpebral fissures as well as high arched palate and denervation atrophy of the muscle which challenges the anaesthesiologist while securing airway and controlling muscle relaxation.[18,19]

Hallermann-Streiff Syndrome is characterized with midfacial hypoplasia, microcornea, mandibular hypoplasia, beaked nose and dwarfism. Every patient with Hallermann-Streiff Syndrome has congenital cataract and may present at the operation room for cataract extraction. The patients with this syndrome have narrow upper airway associated due to craniofacial configuration.[20] This feature as well as obstructive sleep apnoea complicates anaesthetic management and requires further attention.[21]

Patients with Fetal Alcohol Syndrome are exposed to large amounts of alcohol during the first trimester. They have facial abnormalities, low birth weight, developmental and mental retardation, optic nerve hypoplasia or atrophy.[22] They frequently have a cardiac anomaly such as Fallot tetralogy and early mortality is highly due to cardiac disorders and pulmonary infection. They may require surgery due to strabismus, ptosis and anomalies of anterior segment. Difficult airway management is the main problem of these patients during anaesthetic management.[23]

In conclusion, paediatric syndromic patients with craniofacial abnormalities and ophthalmic manifestations need attentive perioperative care when anesthetic management is employed. The anesthesiologist should suspect a related condition and demand further evaluation to confirm the diagnosis in these patients since particular preoperative preparations are required in each syndrome. However, the risk of difficult airway management is the common issue in every patient with craniofacial anomalies.

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There are no conflicts of interest.

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