Orofacial Aspects Of Young Patient With Apert Syndrome

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Apert syndrome is rare congenital disorder with autosomal dominant mode of transmission [1]. In 1906, Franch pediatrician Eugene Apert first described this congenital disorder and his name is associated with the syndrome. Phenotypic manifestations of the disease are explained by premature fusion of cranial sutures. Apert syndrome is the rare acrocephalosyndactyly syndrome type 1 with changes on skull, face, mouth, hands and feet. Apert syndrome belongs to craniosynostosis, because abnormalities of the bones of the skull are the result of premature or intra-uterine adhesion of: coronal, sphenoparietal, squamous and other sutures. Therefore serious changes in the skull and facial bones incurred. Because of early intrauterine adhesion of sutures growth and shape of the skull is disturbed: the skull is high and its tip is sharpened (oxycephaly) and severe asymmetric. Due to the reduced growth of the skull, due to its premature healing, on the one hand, and the expansion of the brain, on the other hand, the inner surface of the skull bones are developing digital impressions, prints brain turns. Orbital gaps are lagging behind in their growth, they are shallow, and because of that pronounced exophthalmos develops. Sometimes exophthalmos is so intense that children can not close eyelids, leading to damage to the bulge of the eye (bulbus oculi) and occurrence of blindness is also possible. Expressed hipertelorism is an abnormally increased distance between orbits (increase distance between eyes (one from another). The nose is small, short, broad and saddle.

The upper jaw is also lagging behind in growth, in transverse and sagittal direction, because it is attached to the base of thread skull by sutures. Narrow maxillary arch, dental crowding and ectopia, high gothic palate, cleft palate in about 25% of the cases and pseudoprognathism exist. The person is always with asymmetrical face, low hairline, webbed neck, pectus excavatum and severe bilateral syndactyly of hands and feet. There is a considerable degree of mental retardation. Changes in the hands and feet are always present. Syndactyly is an abnormally merging 2.3 and 4 fingers and toes.

The aim of Wood’s retrospective study was to quantify the level of dental developmental delay in a group of patients with Aperts syndrome when compared to matched controls. Twenty-six Dental Panoramic Tomographic (DPT) radiographs of patients with Apert syndrome attending Great Ormond Street Hospital were compared to controls (n = 29) from the Eastman Dental Hospital, UK. Dental age, as estimated using the 12 stages of Haavikko and eight stages of Demirjian, suggested no statistical evidence of developmental delay between the Aperts and control group in Wood E et al study [2].

In the Carpentier S et al. report, they demonstrated that a combined orthodontic and orthognathic surgical treatment plan could significantly improve the occlusal function as well as the facial and occlusal aesthetics in patients with Apert syndrome [3].

Kunwar F in his study points out that human genetic diseases needs differential diagnosis to optimize clinical management, enable prenatal detection and genetic counseling. Kunwar F case study shows the importance of

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Phenomizer and molecular genetic analysis in differential diagnosis of genetic diseases [4]. Treating patients with Apert syndrome, Fadda MT et al divide treatment plan in three steps: step 1-from birth to age 2, step 2-growth period, step 3-adult age. They also suggest orthodontic presurgical treatment and tailored surgery timing, scheduling, combining and coordinating actions have to be taken at different stages of the patient’s age, with the aim to reduce the number of general anaesthesia and thus simplifying therapy for both Apert patients and their families [5]. Nasotracheal intubation for a patients with Apert syndrome can be challenge, because of abnormal facial anatomy. Often patients with Apert syndrome underwent partial resection of mandible and cleft palate repair with nasotracheal intubation. Tsukamoto M et al during conduction of nasotracheal intubation used a gastric tube and extubation using an airway exchange catheter [6].

Oral symptoms are explained by decrease in especially anteroposterior diameter of the maxilla with resultant crowding of teeth, and increase in anterior opening of the oral cavity.

Multidisciplinary team include dentist, neurosurgeon, plastic surgeon, physiatrist, ophthalmologist, perinatologist and geneticist are essential for successful management.

Reference


