



## NARRATIVE REVIEW

### Craniofacial Abnormalities and Syndromes

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#### ABSTRACT

Symmetry and proportion plays an important role in cosmetic part of face and golden ratio depicts a strong correlation between mathematical proportion of face and appearance. This can be affected by multiple factors including syndromes which contribute a significant role in craniofacial abnormalities. This paper intends to bring out the craniofacial manifestations of syndromes which play an essential role in diagnosis and more interestingly serve as a window to overall systemic health exploring the associated systemic manifestations.

*Key words: craniofacial abnormalities, diagnosis, syndromes*

An ideal beauty often involves the interpretation of some entity as being in balance and harmony with nature. The earliest Western theory of beauty can be found in the works of early Greek philosophers from the pre-Socratic period, such as Pythagoras. The Pythagorean School saw a strong connection between mathematics and beauty. In particular, they noted that objects proportioned according to the golden ratio seemed more attractive. Ancient Greek architecture is based on this view of symmetry and proportion. Modern research also suggests that people whose facial features are symmetric and proportioned according to the golden ratio (1.618) are considered more attractive than those whose faces are not. Symmetry is also important because it suggests the absence of genetic or acquired defects. Although style and fashion vary widely, cross-cultural research has found a variety of commonalities in people's perception of beauty. Large eyes and a clear complexion, for example,

are considered beautiful in both men and women in all cultures. Neonatal features are inherently attractive and youthfulness in general is associated with beauty. However when the symmetry and proportion are not in balance and harmony with nature then disfigurement results which may be due to various causes including syndromes which contribute an important reason for loss of beauty<sup>(1,2)</sup> The term syndrome has been applied to collection of signs, to groups of symptoms, and to mixed assortments of signs and symptoms.<sup>(3)</sup> Craniofacial manifestations are a central aspect of aesthetic aspect of a syndrome among the multitude of major associated pathologies. The following syndromes are a part of such prominent pathologies portraying the pathetic craniofacial manifestations.

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**Craniofacial Abnormalities of Syndromes****A-APERT'S SYNDROME  
(Acrocephalosyndactyly)**

It is a rare syndrome transmitted by an autosomal dominant Gene which was mentioned as early as 1842 by Baumgartner and Wheaton in 1894. However the eponymic credit is given to Apert for his presentation of the syndrome in 1906. The cause remains multifactorial which includes Virus embryopathy following maternal infection, excessive production of cerebrospinal fluid in embryonic life. The cranium has a characteristic oxycephalic appearance with a high prominent steep forehead. The middle third of the face appears flat and underdeveloped producing the relative prognathism. The nose is small and parrot shaped. Hypertelorism, strabismus and proptosis of eyes are noted. <sup>(4,5)</sup>

**B - BOURNEVILLE-PRINGLE SYNDROME  
(Epilepsia, Tuberos sclerosus)**

It is an autosomal dominant condition possibly related to an effect on chromosome 9. It is a neuro cutaneous syndrome which was first recorded by Von Recklinghausen, however Bourneville and Pringle are credited with the classic description of epilepsy, mental deficiency and adenoma sebaceum. The significance of this syndrome is that most patients die before they are 20 years of age but some survive into middle age. Adenoma sebaceum is characteristic of this syndrome. Subungual fibromas are present. Also Intracranial calcifications and seizures are present. <sup>(4,6)</sup>

**C - CROUZON'S SYNDROME (Craniofacial Dysostosis)**

It is an autosomal dominant condition with a suggested etiology that at birth the sutures of the cranial bones were inflamed causing premature closure of fontanelles, early bone synostosis and a latent period of cranial bone growth. It was first

described by Crouzon in 1912. The hypoplastic maxilla gives the patient a frog like appearance. Exophthalmos, Hypertelorism, Hypoplasia of maxilla are present. Large frontal bony swelling is present. <sup>(4,7)</sup>

**D - DOWN'S SYNDROME (Trisomy 21)**

Down syndrome or trisomy 21 is a chromosomal disorder caused by the presence of all or part of an extra 21st chromosome. It is named after John Langdon Down, the British doctor who described the syndrome in 1866. The disorder was identified as a chromosome 21 trisomy by Jerome Lejeune in 1959. The condition is characterized by a combination of major and minor differences in structure. Often Down syndrome is associated with some impairment of cognitive ability and physical growth as well as facial appearance. Down syndrome can be identified during pregnancy or at birth. The craniofacial features include brachycephaly (condition where the head is disproportionately wide), usually small nose associated with a low nasal bridge, small maxilla, more hypoplastic facial middle third and a smaller lower facial third (mandible). An almond shape to the eyes caused by an epicanthic fold of the eyelid with upslanting palpebral fissures. <sup>(4,8)</sup>

**E - EHLERS-DANLOS SYNDROME (Cutis Hyper Elastica, Indian Rubber Man)**

This syndrome was first described in the 17th century by Van Meekeren, however complete description was given by Ehlers-Danlos (Dermatologist) in 1901. It is an autosomal dominant trait common in males. Hyperelastic skin and fragile. After being stretched it returns to its normal position. <sup>(4,9)</sup>

**F - FACIAL-DIGITAL-GENITAL SYNDROME (Aarskog syndrome)**

The Aarskog-Scott syndrome (AAS) is also known as the Aarskog syndrome, faciodigitogenital syndrome, shawl scrotum

syndrome and faciogenital dysplasia. It is a rare disease inherited as autosomal dominant or X-linked and characterized by short stature, facial abnormalities, skeletal and genital anomalies. Downward slant of the eye slits. The patient has a rounded face. There is impairment in the development of the mid section of the face and wide set of eyes. The nose appears small with nostrils tripped forward.<sup>(10)</sup>

#### **G - GOLDENHAAR SYNDROME (Hemifacial microsomia)**

Goldenhar syndrome is a rare hereditary condition characterized by numerous anomalies affecting the first and second branchial arches of the first pharyngeal pouch, the first branchial cleft and the primordia of the temporal bone. The incidence of this condition, also known as oculoauriculovertebral dysplasia or hemifacial microsomia, varies from 1 in 3,500 to 1 in 5,600 live births, and it is present in 1 in 1,000 children with congenital deafness. The male:female ratio of patients is approximately 3:2. Goldenhar syndrome is characterized by abnormalities of the face (hemifacial microsomia, unilateral facial hypoplasia, and lateral facial cleft), eyes (epibulbar dermoid or lipodermoid [mostly bilateral]; colobomas of the upper eyelid, iris, choroidea, and retina; and other eye anomalies), ears (microtia, anotia, preauricular skin tags or blind fistulas, and other external ear malformations) and incomplete development of the nose, soft palate, lip, and mandible.<sup>(11)</sup>

#### **H - HUTCHINSON-GILFORD SYNDROME (Progeria)**

It was first described by Hutchinson & Gilford. It is a combination of Dwarfism, immaturity and Pseudosenility. Because of a peculiar form of hypermetabolism, persons with this affliction succumb to old age and die of coronary disease

during their middle teens. Face is disproportionately small giving the head a hydrocephalic appearance. The ears are small without lobules. The nose is beaked giving a bird facies. Eyebrows and occasionally eyelashes are lost. Scalp hair is lost and replaced by a downy fuzz, giving a newly hatched bird appearance.<sup>(4, 12)</sup>

#### **I -INFANTILE CORTICAL HYPEROSTOSIS (Caffey - Silverman Syndrome)**

It was first described in 1930 by Roske, however the clinical and roentgenographic features was brought to attention by Caffey, Silverman and Smith in 1945. It is an autosomal dominant condition and its etiology has been suggested that it was caused by a congenital anomaly of the vessels supplying the periosteum of the involved bone, the hypoxia effecting a focal necrosis of the overlying soft tissues and resulting in new periosteal bone formation. The average onset of this syndrome is 9 weeks. Symmetrical swelling over the face located over the body and ramus of the mandible is noted.<sup>(4, 13)</sup>

#### **J - JAW WINKING SYNDROME (Pterygoid Levator Synkinesis and Corneomandibular Reflex)**

Marcus Gunn in 1883, described the syndrome as consisting of unilateral congenital ptosis and rapid exaggerated elevation of the ptotic lid of moving the lower jaw to the contra lateral side. The cause is unknown, but it was originally assumed that the syndrome was based on aberrant innervations of the levator palpebrae superioris from the motor branch of the trigeminal because of the close approximation of the nuclei of the third and fifth cranial nerves. However, a supra nuclear, or at least a combined supra nuclear-nuclear, involvement has also been suggested. Ptosis of one eyelid and depression or movement of the jaw to the contralateral side results in opening of the ptotic eyelid.<sup>(4, 14)</sup>

**K - KLIPPEL- FEIL SYNDROME  
(Brevicollis, Congenital Synostosis Of  
Cardiothoracic Vertebrae, Congenital Osseous  
Torticollis)**

It was first described by Klippel and Feil in 1912. It is an autosomal dominant condition more common in females. The suggested etiology is faulty segmentation of the mesodermal somites sometimes between the third and seventh weeks in utero. A defect in maternal intestinal tract and fetal foregut has also been proposed. The syndrome consists of fusion of some, or even all, cervical vertebrae; Shortness of neck, with painless limitation of head movement & low posterior hairline. The whole head seems to sit directly on the thorax, without an interposing neck.<sup>(4, 15)</sup>

**L-LARSEN'S SYNDROME  
( Osteochondrodysplasia )**

The syndrome, first recognised as a distinct entity by Larsen in 1950, includes flattened facies, multiple congenital joint dislocations, clubfoot deformities, and frequently cleft palate. Flattened mid-face, prominent forehead with frontal bossing, depressed nasal bridge are the prominent features.<sup>(16)</sup>

**M - MEDIAN CLEFT FACE SYNDROME  
(Frontonasal dysplasia)**

Median cleft face syndrome is a rare, sporadic condition. It results from embryonic failure of fusion of the median nasal processes. The most common associated malformations are lipoma or agenesis of corpus callosum, tibial hypoplasia, proximal hullucal polydactyly, epibulbar dermoids, ear tags and tetralogy of Fallot. Most patients have a normal intelligence. Anterior cranium bifidum occultum, Ocular hypertelorism, broadening of the nasal root, median cleft nose and a median facial cleft affecting the upper lip and palate, microphthalmia.<sup>(17)</sup>

**N - NAGER'S SYNDROME (Nager acrofacial**

**dysostosis - NAFD )**

Nager syndrome is a congenital malformation syndrome. The prevalence is unknown; more than 100 cases of NAFD have been published. Nager Syndrome is a condition resulting from problems in the development of the first and second branchial arches. NAFD is characterized by mandibulofacial anomalies that include downward slant of palpebral fissures, ptosis of upper lids, coloboma of lower lids, deficiency of eyelashes of the medial one-third to two-thirds of the lower eyelids, hypoplasia of the malar eminences and zygomata, hypoplasia of the maxilla, cleft palate, absence or hypoplasia of the palatal velum, choanal atresia, extension of a "tongue" of temporal hair down the sides of the cheeks. Cleft lip is rare.<sup>(18)</sup>

**O -OROMANDIBULAR LIMB  
HYPOGENESIS SYNDROME ( Hypoglossia-  
hypodactyilia syndrome )**

Oromandibular-limb hypogenesis syndromes (OLHS) represent a group of rare conditions characterized by congenital malformations involving multiple sites such as the tongue, mandible, and limbs. In 1971, Hall classified OLHS into 5 major types. The oromandibular-limb hypogenesis syndrome comprises a group of anomalies which simultaneously affect the mandible, tongue, and maxilla with or without reductive limb anomalies. It is characterized by failure of development of the intraoral region and distal extremities, facial Paralysis, inability to move the eyes from side to side and Crossed eyes (strabismus).<sup>(19)</sup>

**P - PARRY-ROMBERG SYNDROME  
(Progressive Hemifacial Atrophy)**

It is an autosomal dominant condition. It was first described by Parry & Romberg in 1825 & 1846 respectively. The suggested etiology is irritation in the peripheral tropics sympathetic system. It

consists of slowly progressive atrophy of the soft tissues of essentially half the face, accompanied most often by contralateral Jacksonian epilepsy, by trigeminal neuralgia & by changes in hair and eyes. Occasionally there may be associated atrophy of half the body. The body and ramus of the mandible are shorter on the involved side and a delay in development of the angle.<sup>(4, 20)</sup>

#### **R - RUBINSTEIN – TAYBI SYNDROME ( Broad thumb-hallux syndrome )**

It is a genetic disorder associated with mental retardation and physical features such as broad thumbs, toes, short stature and craniofacial abnormalities. The prevalence is approximately 1 case per 300, 000 persons and as high as 1 case per 10,000 live births. The etiology is related to the disruption of the binding protein for cyclic adenosine monophosphate – response element binding protein associated with chromosomal rearrangement. There is downward slant of palpebral fissures, mild hypertelorism, long philtrum, beaked nose, strabismus.<sup>(21)</sup>

#### **S - STURGE WEBER SYNDROME (Encephalotrigeminal Angiomatosis)**

This syndrome was first described by Sturge and Weber in 1897 & 1922. It consists of Venous angioma of leptomeninges overlying the cerebral cortex with ipsilateral angiomatous lesions of face, ipsilateral gyriform calcifications of the brain, epilepsy, mental retardation, ocular involvement and contralateral hemiplegia. Nevus flammeus (portwine stain) commonly occurs on

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the face and is mostly unilateral in the course of trigeminal nerve.<sup>(4, 22)</sup>

#### **T - TREACHER COLLINS' SYNDROME (Mandibulofacial Dysostosis, Franceschetti-Zwahlen-klein Syndrome, Bilateral Facial Agenesis)**

This syndrome was first described by Treacher Collins and Franchetti in 1846 and 1940 respectively. It is an autosomal dominant condition. The suggested etiology is incorrect development of blood relay (from the remains of first aortic arch to stapedia artery to external carotid artery). The main features include downward sloping palpebral fissures, depressed cheek bones, deformed pinna, receding chin, large fish like mouth, tongue shaped process of hair that extends towards cheek.<sup>(4,23)</sup>

#### **W - WHISTLING FACE SYNDROME (Freeman Sheldon syndrome)**

This syndrome was first described by Freeman & Sheldon. The significant features include lips protruding as in whistling, full cheeks with increased philtrum length, small nose and nostrils.<sup>(4, 24)</sup>

#### **CONCLUSION**

The true value of health in relation to appearance and function is felt when pathology causes loss of normal morphology and functions. Syndromes are one such pathology which arises from different etiologies resulting in mild to fatal consequences both with respect to morphology and function. Craniofacial abnormalities are a significant part of syndrome and dental professionals have an essential role to play in diagnosis of syndromes.

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