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Chapter

Diagnosis of Dentofacial Anomolies

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Abstract

It is very challenging to understand and analyse anomalies of dentofacial region. Diagnosis plays a very important role in the further treatment of any condition related to orofacial anomalies. Diagnosis includes taking complete history and required investigations and conclusion. History gives more information towards clinical path, and investigation will lay more emphasis on conclusion. Anomalies involving dentofacial region may be related to tooth, maxilla, mandible, soft tissue anomalies and syndromic conditions. Dentofacial anomolies not only involve the dentofascial region but can spread to various other vital organs, so sometimes correlating the systemic problem will be of prime importance. When the other body is involved, the varied presentation will be a challenge in diagnosis. Multiple organs should be investigated for an diagnostic conclusion. Brining diagnostic information of anomalies is the aim of the chapter. Here, we cover various clinical features, diagnostic criteria, and investigation protocols of dentofacial anomalies.

Keywords: dentofacial anomalies, tooth, syndrome, diagnosis

1. Introduction

Societal forces define norms for an acceptable physical appearance and equate to a good smile. Significant aesthetic and functional issues in both jaws are included in dental abnormalities. It has been discovered that certain syndromes and systemic disturbances are connected with changes in craniofacial form, structure or function to the point where these changes can be categorised as the essential characteristics of such illnesses. It is crucial that the clinician is aware of any dysmorphologic alterations to the orofacial structures in order to consider specific disease entities and rule out the participation of any other tissues or organ systems that might be syndromically connected. Dental appearance with success in life plays an important role. An increased concern for dental appearance has been observed during adolescence and early adulthood. One among them is malocclusion that is described as an irregularity of the teeth or a poor relationship of the dental arches beyond the range of what is accepted as normal. Malocclusion can impact quality of life causing psychosocial limitations (awkwardness in the social context or reduced career opportunities) and functional disturbances (affecting mastication, swallowing and speech; increasing susceptibility to trauma; and increasing prevalence of dental caries, periodontal disease and temporomandibular joint disorders). Even though the malocclusion is not a dentofacial anomalies, it is a part of various dentofacial anomalies.

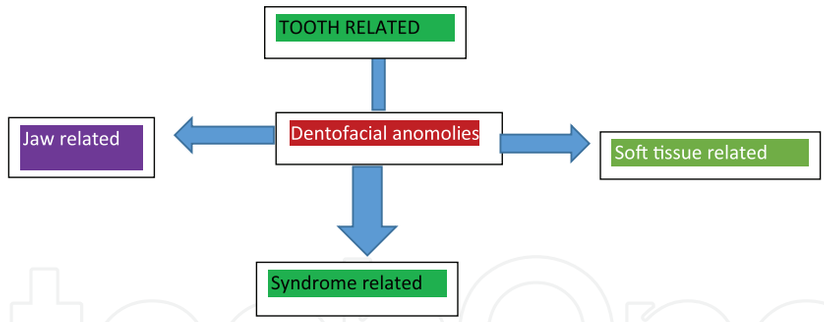

Figure 1.
Distribution of dentofacial anomalies.

For any individual defect, there may be variation in phenotype, associated anomalies and cause. To help organise these various disorders, dysmorphologists have grouped them into 'syndromes', 'sequences' and 'associations' based on our level of understanding of their aetiologies as shown in **Figure 1**.

2. Developmental anomalies of teeth

In the course of their lifetime, humans produce two sets of teeth: the primary dentition and the permanent dentition. By the age of 12, the primary dentition is fully replaced by permanent teeth that last for lifetime, as opposed to many animals, which have numerous sets of teeth that erupt depending on how and when a functional tooth is used and exfoliated.

Human teeth begin to develop during foetal development and continue growing until 10 years after birth. Teeth abnormalities develop for a number of reasons, such as poor nutrition, systemic illnesses, genetic problems, that influence a person throughout this time.

The developmental disturbances of teeth can be classified as anomalies affecting the following features:

- size
- shape
- number
- surface disturbances

2.1 Microdontia

When teeth are physically smaller than usual, the term 'microdontia' should be used. As one or more developing lobes of a tooth germ fail, resulting in microdonts, the condition can also cause aberrant form.

Both generalised and localised microdontia exist. Generalised microdontia is typically linked to a growth hormone or pituitary dysfunction-related developmental disorder. A single tooth in the arch is typically affected by localised microdontia. The maxillary lateral incisors (peg laterals) and third molars are the most commonly afflicted teeth. Usually, a size comparison between the neighbouring and opposing teeth makes the diagnosis simple.

2.2 Macrodontia

Megadontia or megalodontia are other names for macrodontia. The phrase is used when one or more teeth stand out as being larger than they should be. When present in a normal person, macrodontia causes crowding and malocclusion. However, the entire dentition may be impacted without occlusal inconsistencies in cases of growth of hormone-related gigantism [1, 2]. True-generalised macrodontia, as this condition is known, is incredibly uncommon. Relative macrodontia is a condition that is frequently observed in which the teeth may be slightly larger in size or may appear huge because of a smaller jaw size. A single tooth's macrodontia may be caused *via* fusion or gemination.

Patients with macrodontia frequently have aesthetic concerns, and the diagnosis is aided by clinically bigger teeth and malocclusion. Normal internal tooth structures are visible on radiographs**.**

3. Developmental disturbances in shape of teeth

3.1 Gemination

Gemination is an aberration that develops when a single tooth germ tries to divide by invaginating, which results in the partial creation of two teeth. The tooth typically has one or two crowns, which may be totally or partially separated, but only one root and one root canal [1].

Clinically, a single root has a bifid crown with complete segregation or minor grooving between two big crowns. The total number of teeth stays the same during gemination because only a single tooth bud can be partially split to change the number of teeth. The most often impacted teeth in both dentitions are the incisors and canines.

These are common pulp canals and either a single or partially divided pulp chamber, according to radiology, which are greater than usual crown width with a shallow groove (**Figure 2**).

3.2 Fusion

Fusion is the joining of two distinct tooth germs, which can be seen radiographically as two different pulp chambers and root canals [2]. The action of pressure or mechanical physical force that produces close contact between two erupting teeth

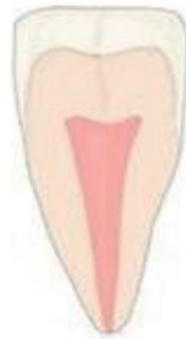

Figure 2.
Features of gemination.

Figure 3.
Courtesy: JSS Dental College, figure showing fusion of lateral incisor with supernumerary tooth.

was identified as a possible explanation, although the exact cause of fusion is still unknown. It occurs more frequently in teeth at the front. In contrast to permanent teeth, it occurs more frequently in deciduous teeth [3].

Clinical: With or without a bifid crown, the tooth is about twice as large as it should be. Root canals in a tooth may be single or combined. Fusion may happen between a normal and a supernumerary tooth or result in fewer teeth [3, 4]. The union's true character and scope will become increasingly clear (**Figure 3**).

3.3 Concrescence

Concrescence is two fully formed teeth, joined along the root surfaces by cementum. The process is noted more frequently in the posterior and maxillary regions. Lack of adequate space and crowding of teeth are the most accepted elucidated aetiology.

Diagnosis is made commonly with the help of radiographs. It is not always possible to distinguish among concrescence, teeth in close contact and superimposed teeth. Radiograph will show union of two teeth with the help of cementum.

3.4 Talon's Cusp

The talon cusp, an aberrant structure that resembles an eagle's talon, is produced by the cingulum regions of a maxillary or mandibular permanent incisor [4], commonly observed on the lateral or central maxillary incisor.

Clinical: A T-shaped elevation on the tooth makes Talon's cusp an easy diagnosis.

Radiological: Where it occurs, overlaid on the incisors, there is a coating of enamel that appears to be normal, and the outline is smooth.

3.5 Dilaceration

The term 'dilaceration' refers to an angulation, or a sharp bend or curve, in the root or crown of a formed tooth.

Clinical diagnosis is not possible.

Radiological: Curve or bending occurs anywhere along the length of tooth, sometimes at cervical portion or midway along the root or even just at the apex of root.

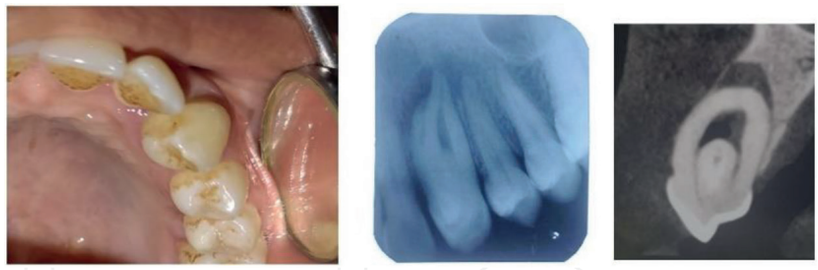

Figure 4.
Clinical and intra-oral radiographic presentation of the condition. Courtesy. JSS Dental College.

3.6 Dens in dente (dens invaginatus)

The 'dens in dente' is a developmental variant that is believed to come from an invagination in the tooth crown's surface prior to calcification. Prior to the calcification of the dental tissues, there is histologically observed deepening or invagination of the enamel organ into the dental papilla [5].

Clinical: The tooth's labial face is frequently bulbous. Conical or asymmetrical crown shapes are both possible. Dental infolding is a rare clinical occurrence. Rarely, primary teeth may be impacted.

Radiological: It can be identified as a pear-shaped invagination of enamel and dentin that has a close resemblance to the pulp in depth and a tight constriction at the aperture on the surface of the tooth. The tooth appears to be 'inverted' (**Figure 4**).

3.7 Dens evaginatus

The dens evaginatus is a developmental condition that appears clinically as an accessory cusp or a globule of enamel on the occlusal surface between the buccal and lingual cusps of premolars, unilaterally or bilaterally, although it has been reported to occur rarely on molars, cuspids and incisors.

Schulze [6] distinguished the following five types of DE for posterior teeth by the location of the tubercle.

1. A cone-like enlargement of the lingual cusp.

2. A tubercle on the inclined plane of the lingual cusp.

3. A cone-like enlargement of the buccal cusp.

4. A tubercle on the inclined plane of the buccal cusp.

5. A tubercle arising from the occlusal surface obliterating the central groove.

Clinical: It appears as a tubercle of enamel on occlusal surface of the affected tooth. Polyp-like protuberance in central groove on lingual ridge of buccal cusp is seen.

Radiological: Occlusal surface has tuberculated appearance.

3.8 Enamel pearl

Heterotopic presence of enamel in the form of a globule is called enamel pearl. It is usually found on the root surface.

Clinical: It appears as a yellowish white, spherical structure adherent to the furcation area of the root surface. The diameter ranges from 1 mm to 3 mm.

Radiological: It appears as smooth, round and well-defined radiopacity present along the root surface. Radiodensity is same as that of the enamel.

3.9 Taurodontism

It is characterised by clinical and anatomical crown of normal shape and size, an elongated body and short roots with longitudinally enlarged pulp chambers [7].

Clinical: Affected teeth tend to be rectangular and exhibit pulp chambers with a dramatically increased apico-occlusal height and a bifurcation close to the apex [7, 8].

Radiological: Pulp chamber is extremely large with much greater apico-occlusal height than normal. Extensions of rectangular pulp chamber occur into elongated body of the tooth [7]. Pulp lacks the usual constriction at the cervix of tooth. The root and root canals are exceedingly short. There is also increased dimension between cementoenamel junction and furcation.

3.10 Supernumerary roots

Teeth that are normally single-rooted exhibit two roots.

Clinical: They develop as slender outgrowths at the centre of furcation area of molar teeth.

Radiological: If the bifurcation produces two distinct apices and these are arranged as one mesial to the other, then it will be seen on the radiographs. If the two apices are on the labial and lingual side, they may get superimposed on each other appearing as a bulbous root, which may mimic hypercementosis.

4. Developmental disturbances in structure of teeth

Amelogenesis imperfect: A complex collection of diseases known as amelogenesis imperfecta shows developmental changes in the enamel's structure when no underlying systemic problem is present.

Inaccuracies in hypoplastic amelogenesis.

Pits that range in size from a pinhead to a pea are dispersed around the teeth's surface. The pits can be placed in rows or columns and are more noticeable on the buccal surfaces of the teeth.

Localised pattern: Linear depressions and horizontal rows of pits can be seen on the affected teeth. The affected region is typically found in the middle third of the buccal surfaces of the teeth. Typically, neither the incisal edge nor the occlusal surface is impacted.

All teeth have an enamel that is thin, firm, shiny, and has an autosomal dominant smooth pattern. There is lack of the proper enamel thickness.

Radiographs exhibit a thin peripheral outline of radiodense enamel. Unerupted teeth, often undergoing resorption, may be seen.

Enamel agenesis: A total lack of enamel formation. The teeth are the shape and colour of the dentin, with a yellow-brown hue, open contact points and crowns that taper towards the incisal-occlusal surface. The surface of the dentin is rough, and an anterior open bite is seen frequently.

Radiographs demonstrate no peripheral enamel overlying the dentin.

4.1 Hypomaturation amelogenesis imperfecta

Pigmented pattern: The surface enamel is mottled and agar brown. The enamel often fractures from the underlying dentin and is soft enough to be punctured by a dental explorer.

X-linked pattern: The deciduous teeth are opaque white with a translucent mottling; the permanent teeth are opaque yellow-white and may darken with age. Focal areas of brown discoloration may develop within the white opaque enamel [9].

Snow-capped patterns: A zone of white opaque enamel on the incisal or occlusal one quarter to one-third of the crown.

4.2 Hypocalcified amelogenesis imperfecta

On radiographs, the teeth show a thin radiopaque enamel outline around the periphery. Unruptured teeth showing signs of resorption are common.

Both dentitions have diffuse thin, smooth and glossy enamel in an X-linked pattern. Open contact points and crown preparation shapes are common in teeth. Brown to golden brown is the range of colour.

An outline of radiopaque enamel can be seen on radiographs.

Rough surface: The enamel is thin and firm and has a rough pattern. Similar to the smooth forms, the teeth have open contact sites and taper towards the incisal-occlusal surface. From white to bright white, the colour varies.

Clinical: On eruption, the enamel is yellow-brown or orange, but it often becomes stained brown to black and exhibits rapid calculus apposition.

A thin radiopaque enamel outline around the teeth's periphery can be seen on radiographs. Unruptured teeth that are showing resorption are common.

Thin, shiny, smooth enamel is diffused in both dentitions with an X-linked pattern. There are exposed contact sites and the teeth frequently resemble crown preparations. Brown to yellow-brown are the different shades.

Enamel that is radiopaque can be seen around the edges on radiographs.

The enamel has a rough surface and is thin and firm. The teeth display open contact points and taper towards the incisal-occlusal surface just like in the smooth forms. White to yellow-white can be seen throughout the spectrum.

Radiological: the density of the enamel and dentin are similar. Before eruption the teeth are normal in shape; however, after a period of function much of the cuspal enamel is lost, with the occlusal surface becoming the most irregular (**Figure 5**) [10].

4.3 Dentinogenesis imperfecta

Both deciduous and permanent teeth are affected by the autosomal dominant syndrome known as dentinegenesis imperfecta [11, 12].

Clinical: Affected teeth have large crowns, grey to yellowish brown colour, and constricted cervical areas give them a 'tulip' form.

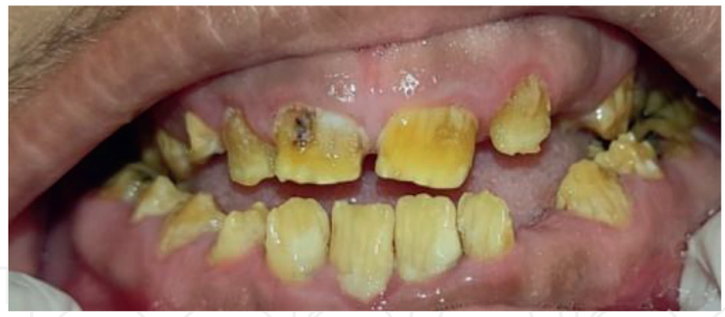

Figure 5.
Courtesy: JSS Dental College, clinical picture of amelogenesis imperfecta.

Radiologically, the teeth appear to be solid and devoid of root canals and pulp chambers. Because enamel is easily fractured, exposed dentin has rapid attrition. The teeth feature narrow roots, bulbous crowns, cervical constriction, and early pulp chamber and root canal obliteration.

Dentition with enamel that is normal in thickness, dentin that is incredibly thin and pulps that are noticeably enlarged is dentin dysplasia.

It is a rare disturbance of dentin formation, characterised by normal but atypical dentin formation, with abnormal pulp morphology.

Clinical:

Type I (radicular). Both dentitions are affected, although the teeth appear clinically normal in morphologic appearance and colour. Occasionally, there may be a slight amber translucency. However, the teeth characteristically exhibit extreme mobility and are commonly exfoliated prematurely or after only minor trauma as a result of their abnormally short roots.

Type II (coronal): The deciduous teeth have the same yellow, brown or bluish-grey opalescent appearance as seen in dentinogenesis imperfect [12].

Radiological:

Type I (radicular): In both dentitions, the roots are short, blunt, conical or similarly malformed. In the deciduous teeth, the pulp chambers and root canals are usually completely obliterated, while in the permanent dentition, a crescent-shaped pulpal remnant may still be seen in the pulp chamber.

Type II (coronal): Bulbous crowns, cervical constriction, thin roots, and early obliteration of the pulp. The permanent teeth demonstrate normal clinical coloration; however, radiographically, the pulp chambers exhibit significant enlargement and apical extension. This altered pulpal anatomy has been described as thistle tube shaped or flame shaped (**Figure 6**).

4.4 Regional odontodysplasia

Regional odontodysplasia is a specific, non-hereditary anomaly of tooth development that has severe negative effects on the growth of enamel, dentin and pulp [13].

Clinical

Asymmetrical in shape appears yellow to brownish and is often tiny. They either indicate a delay in eruption or a complete failure [12.14]. They have a noticeably different shape, are typically quite uneven in appearance and frequently show signs of poor mineralisation.

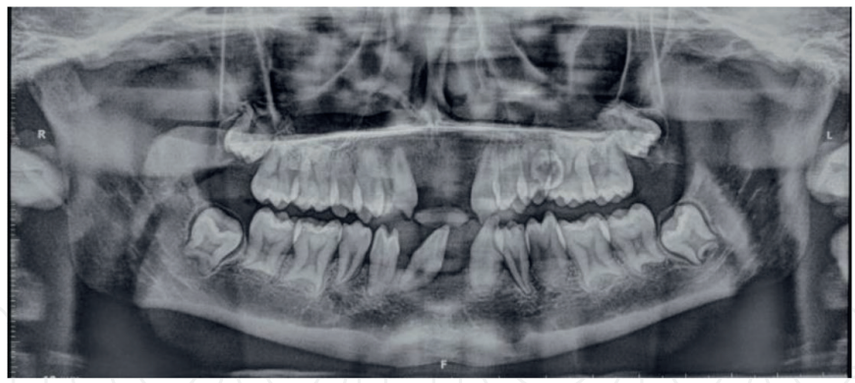

Figure 6.
Courtesy: JSS Dental College. Radiograph showing malformed tooth with short roots in Type I dentin dysplasia.

Radiological

The nickname 'ghost teeth' refers to the changed teeth's light, wispy appearance caused by the presence of radiolucent pulp surrounded by extremely thin enamel and dentin. There is little contrast between the dentin and the enamel, and the coronal silhouette is blurry or unclear. There may be visible short roots and open apices. The thickened pulps frequently show one [14].

5. CT image of cleft palate, courtesy JSS Medical College, Mysore

5.1 Soft tissue anomolies

5.1.1 Cleft lip and palate

Birth deformities such as cleft lip and palate can cause a person to have a number of orofacial malformations. One of the most prevalent birth defects, orofacial clefts can occur alone or in combination with other congenital malformations. Both syndromic and non-syndromic clefts with accompanying abnormalities make up a sizable portion of these clefts [15, 16].

Openings or cracks in the upper lip, the palate or both are known as cleft lip and cleft palate, respectively. When a developing baby's facial tissues do not fully seal, it can lead to cleft lip and cleft palate. The most prevalent birth malformations are cleft lip and cleft palate. Although they most frequently manifest as solitary birth abnormalities, they are also linked to a variety of inherited genetic diseases or syndromes (**Figure 7**).

Diagnostic features:

Typically, a split (cleft) in the lip or palate is obvious from birth. Cleft lip and palate can manifest as follows:

a facial split that affects one or both sides of the lip and palate (roof of the mouth).

a break in the lip that is only visible as a tiny notch or that extends through the upper gum and palate and into the base of the nose. A crack in the roof of the mouth that has no impact on how the face looks. Less frequently, a cleft only affects the soft palate muscles in the rear of the mouth, where the lining of the mouth covers them (submucous cleft palate). This kind of cleft is common [16].

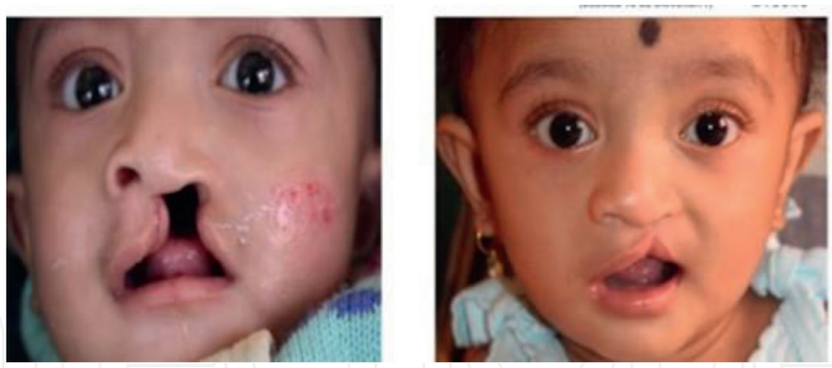

Figure 7.
Baby with cleft lip and palate deformity, showing corrected cleft palate and lip (photos courtesy JSS Dental College, cleft centre).

Signs and symptoms of submucous cleft palate may include the following:

1. Difficulty with feedings.

2. Difficulty swallowing, with potential for liquids or foods to come out the nose.

3. Nasal-speaking voice.

4. Chronic ear infections Diagnosis.

Most cases of cleft lip and cleft palate are noticed right away at birth and do not require special tests for diagnosis. Increasingly, cleft lip and cleft palate are seen on ultrasound before the baby is born (**Figures 8** and **9**).

Ultrasound before birth

During a prenatal ultrasound, sound waves are used to produce images of the growing foetus. A doctor may notice a variation in the face structures after reviewing the images.

Beginning about the 13th week of pregnancy, ultrasonography can identify cleft lip. Accurately diagnosing a cleft lip may get simpler as the foetus continues to

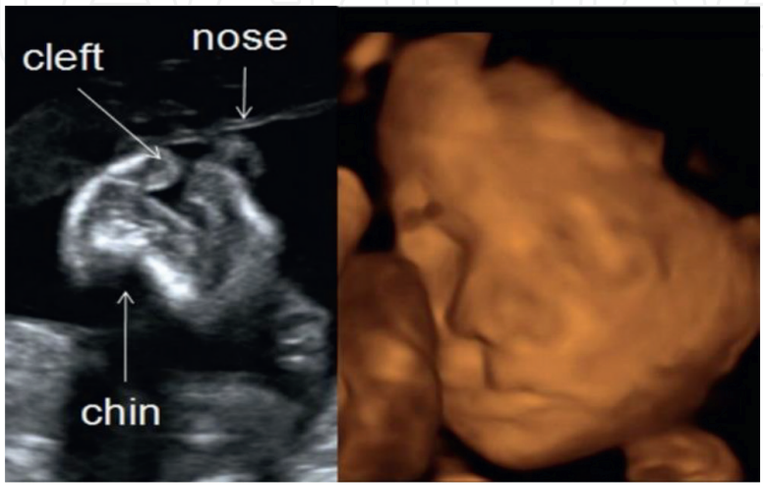

Figure 8.
OPG showing the bony defect in the upper anterior with irregularly arrenged to tooth. Courtesy JSS Dental College, Mysore.

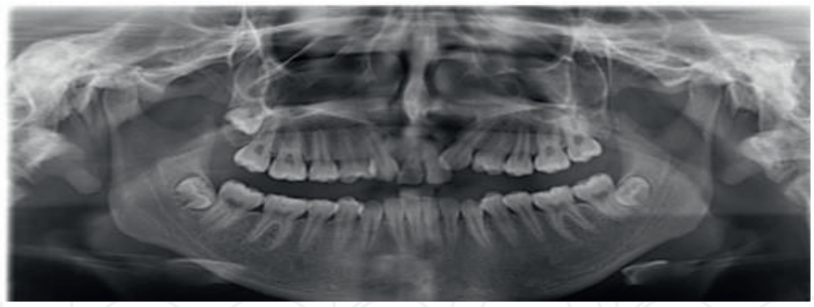

Figure 9.
CT image of cleft palate, courtesy JSS Medical College, Mysore.

develop. When a cleft palate develops on its own, ultrasonography imaging is more challenging (**Figure 10**).

Your doctor might suggest a treatment to remove a sample of amniotic fluid from your uterus if a prenatal ultrasound reveals a cleft (amniocentesis). The fluid test could reveal a genetic condition that could lead to other birth abnormalities in the foetus. But the most common reason for cleft lip and cleft palate is shown **Table 1**.

5.2 Syndromes

5.2.1 Downs syndrome

A second whole or partial copy of chromosome 21 is produced as a result of faulty cell division, which results in the genetic condition known as Downs syndrome. Downs syndrome's physical characteristics and developmental abnormalities are brought on by this excess genetic material.

Individuals with Downs syndrome may have varying degrees of intellectual disability and developmental delays. It is the most prevalent genetic chromosomal defect and the root of children's learning problems. It frequently results in other medical

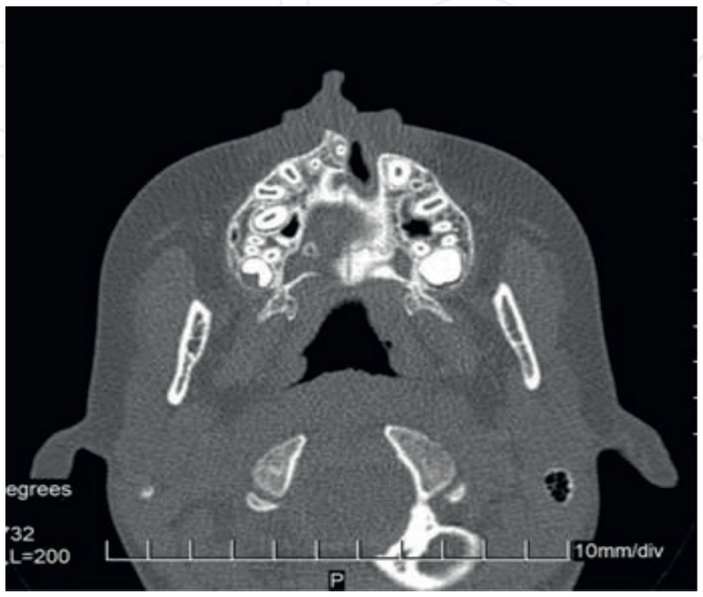

Figure 10.
Fetoscopic image of cleft lip—courtesy: JSS MEDICAL COLLEGE, MYSORE.

Associated syndromes with cleft lip and palate
Autosomal dominant syndromes
Apert
Cleidocranial dysostosis
Hay-Wells
Treacher Collins
Vander Woude
Oculodentodigital
Autosomal recessive syndromes
Cerebro-costo-mandibular
Dubowitz
Mohr
Robert
X-linked inheritance
Oro-facial-digital
Oto-palato-digital
Chromosomal disorders
Mutation in 3p arm, 5p arm, 9p arm and 18q arm
Trisomy 4p, 9p
Trisomy 13
Trisomy 18

Table 1.
Syndrome associated with cleft lip and palate.

issues as well, such as cardiac and gastrointestinal problems. With Downs syndrome, both children and adults have distinctive face features (**Figure 11**) [17].

Though not all people with Downs syndrome have the same features, some of the more common features include the following:

Flattened face
Small head
Short neck
Protruding tongue
Upward slanting eye lids (palpebral fissures)
Unusually shaped or small ears
Poor muscle tone
Broad, short hands with a single crease in the palm
Relatively short fingers and small hands and feet
Excessive flexibility
Tiny white spots on the coloured part (iris) of the eye called Brushfield's spots.
Short height

This extra genetic material is responsible for the characteristic features and developmental problems of Downs syndrome. Any one of three genetic variations can cause Downs syndrome:

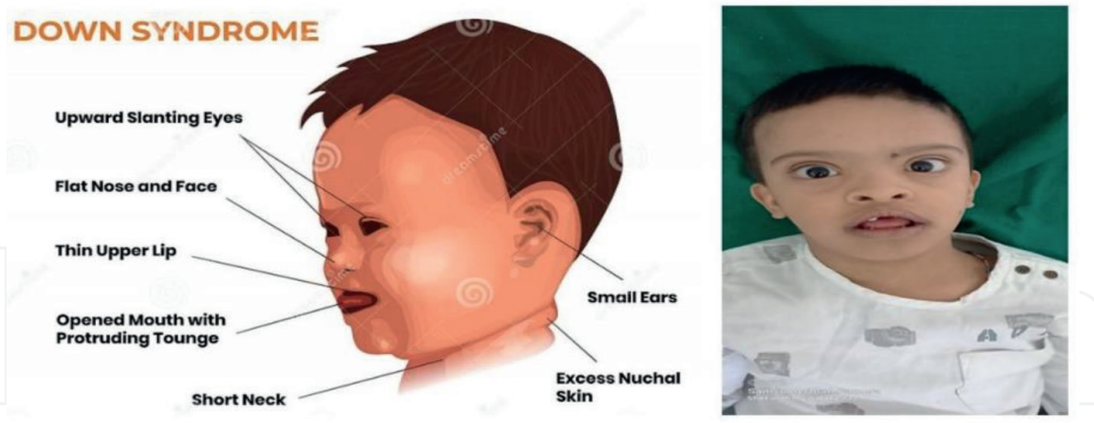

Figure 11.
Salient features of Downs syndrome.

Trisomy 21. About 95 percent of the time, Downs syndrome is caused by trisomy 21—the person has three copies of chromosome 21, instead of the usual two copies, in all cells [18]. This is caused by abnormal cell division during the development of the sperm cell or the egg cell.

Mosaic Downs syndrome. In this rare form of Downs syndrome, a person has only some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilisation.

Screening tests during pregnancy

Screening for Downs syndrome is offered as a routine part of prenatal care. Although screening tests can only identify your risk of carrying a baby with Downs syndrome, they can help you make decisions about more-specific diagnostic tests.

Screening tests include the first trimester combined test and the integrated screening test.

The first trimester combined test, which is done in two steps, includes the following:

Blood test. This blood test measures the levels of pregnancy-associated plasma protein-A (PAPP-A) and the pregnancy hormone known as human chorionic gonadotropin (HCG). Abnormal levels of PAPP-A and HCG may indicate a problem with the baby [19].

Nuchal translucency test. During this test, an ultrasound is used to measure a specific area on the back of your baby's neck. This is known as a nuchal translucency screening test. When abnormalities are present, more fluid than usual tends to collect in this neck tissue.

Using your age and the results of the blood test and the ultrasound, your doctor or genetic counsellor can estimate your risk of having a baby with Downs syndrome.

Integrated screening test

During the first and second trimesters of pregnancy, the integrated screening test is administered in two parts. To calculate the likelihood that your child has Downs syndrome, the findings are pooled.

Initial trimester. An ultrasound is used in part one to measure nuchal translucency and a blood test to measure PAPP-A.

First trimester. Alpha fetoprotein, estriol, HCG, and inhibin A are the four pregnancy-related chemicals that are measured by the quad screen in your blood.

Pregnant women's diagnostic procedures

Consider additional testing to confirm the diagnosis if your screening test results are positive or concerning, or if you have a high risk of having a baby with Downs syndrome. You can balance the benefits and drawbacks of these tests with the aid of your healthcare provider.

Diagnostic tests that can identify Downs syndrome include the following:

Sample chorionic villus (CVS). Cells from the placenta are utilised in CVS to examine the foetal chromosomes. Between 10 and 13 weeks of pregnancy, the first trimester is the traditional time for this test to be carried out. A CVS carries a very minimal chance of pregnancy loss (miscarriage).

Amniocentesis. A needle is introduced into the mother's uterus to remove a sample of the amniotic fluid around the foetus. The chromosomes of the foetus are then examined using this sample. After 15 weeks of pregnancy, doctors typically administer this test in the second trimester. A very small risk of miscarriage is also associated with this test.

For couples undergoing *in vitro* fertilisation who are at heightened risk of passing along specific genetic traits, preimplantation genetic diagnosis is a possibility.

Diagnostic tests for newborns

Initial Downs syndrome diagnoses are frequently made based on a baby's looks after delivery. However, babies without Downs syndrome can also have the characteristics linked with the condition, so your doctor will likely request a test called a chromosomal karyotype to confirm the diagnosis. This test examines your child's chromosomes using a blood sample. Downs syndrome is the result of an extra copy of chromosome 21 in all or some cells.

Translocating the Downs syndrome. Additionally, Downs syndrome can develop before or during conception if a piece of chromosome 21 translocates (attaches to another chromosome). These kids have two copies of chromosome 21 as usual, but they also contain additional chromosome 21 genetic material linked to another chromosome.

5.3 Crouzon Syndrome

Synonyms include the following:

craniofacial dysostosis,

craniostenosis, Crouzon type,

Crouzon craniofacial dysostosis.

Crouzon syndrome is a rare genetic disorder. It is a form of craniosynostosis, a condition in which there is premature fusion of the fibrous joints (sutures) between certain bones of the skull. Symptoms primarily include abnormalities of the face and head [20].

Signs and symptoms

The primary features of Crouzon syndrome, also known as craniofacial dysostosis, are pronounced facial deformities and premature closure of the fibrous joints (cranial sutures) connecting some of the skull's bones. Malformations of the cranium and face can range from minor to potentially severe, even in members of the same family who are typically unaffected [21]. Crouzon syndrome is inherited autosomally dominantly and is brought on by changes (mutations) in one of the FGFR genes, typically FGFR2.

The bony cavities of the skull that house the eyeballs or the orbits in the majority of people are unusually shallow. The consequence is a protrusion or bulge forward of the eyeballs, known as proptosis [22]. About 30% of those with Crouzon syndrome go on to develop hydrocephalus, a disorder marked by reduced flow.

Diagnosis

Based on a thorough clinical evaluation, the recognition of recognisable physical characteristics and a battery of specialised testing, Crouzon syndrome is typically diagnosed at birth or during infancy. Advanced imaging methods, such as magnetic resonance imaging (MRI) or computed tomography (CT) scanning, may be used during such testing.

Clinical evaluation and testing

MRIs and CT scans are utilised to identify or detect some abnormalities that may be connected to the illness (e.g. craniosynostosis, other skeletal abnormalities). X-rays and a computer are used in CT scanning to produce a film that shows cross-sectional images of inside structures. A magnetic field and radio waves are used in MRI to produce finely detailed cross-sectional images of certain organs and tissues.

A Crouzon syndrome diagnosis can be confirmed by molecular genetic testing (**Figures 12** and **13**).

5.4 Foetal alcohol syndrome

This is the most severe foetal alcohol spectrum disorder. These are a group of birth defects that can happen when a pregnant woman drinks alcohol (**Figure 14**). Other foetal alcohol syndrome disorders (FASDs) include the following [22, 23]:

partial foetal alcohol syndrome,
alcohol-related birth defects,
alcohol-related neurodevelopment disorder,
neurobehavioural disorder associated with prenatal alcohol exposure.

They can include the following:

problems with the heart, kidney and bones,

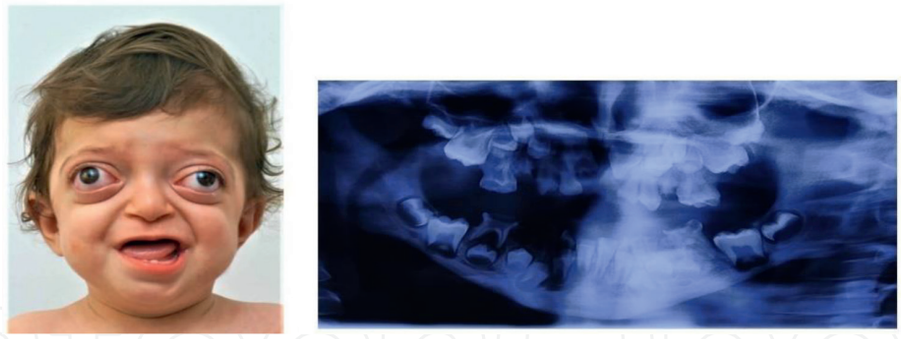

Figure 12.
Salient features of Crouzon syndrome and panoramic image to show bone density and affected teeth.

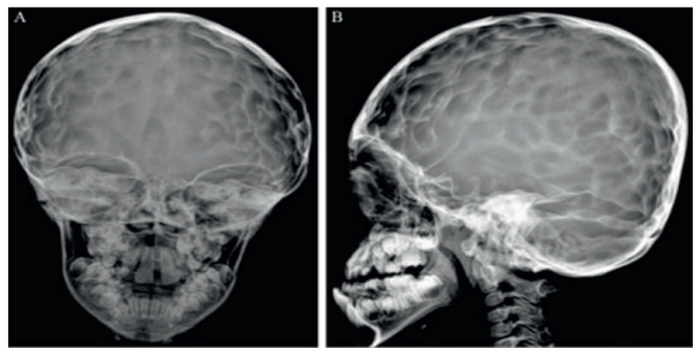

Figure 13.
Skull radiograph A, anterior-posterior and B, lateral views showing prominent convolutional markings appearing as a copper beaten skull. Courtesy: JSS Medical College, Mysore.

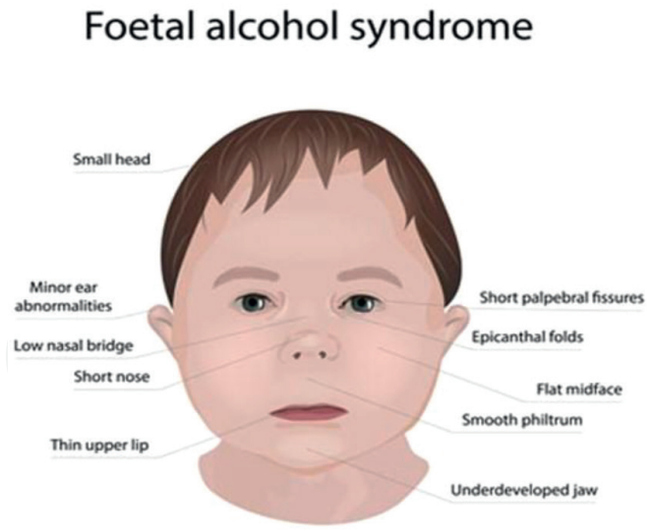

Figure 14.
Salient featues of foetal alcohol syndrome.

learning disabilities and low IQ,
trouble with memory, coordination and attention,
hyperactivity.

Problems with sleep and suckling as an infant. Foetal alcohol syndrome can have many symptoms, including the following:

Physical defects:
small head and brain size,
vision or hearing problems,
joint, limb and finger deformities,
distinctive facial features such as small eyes, thin upper lip and a ridge between the nose and upper lip.

Neurological problems:
learning problems,
coordination and balance problems,
trouble reasoning,
hyperactivity,
moodiness.

Behavioural issues:
poor social skills,
difficulty in school,
poor impulse control.

5.5 Goldenhar syndrome (GS)

Oculoauriculovertebral dysplasia, also referred to as hemifacial microsomia, manifests clinically in a variety of ways, including abnormalities of the craniofacial, vertebral, cardiac, renal, and central nervous systems1. Epibulbar dermoids, microtia, mandibular hypoplasia, and spinal abnormalities are among the symptoms of GS that are frequently present. Hemifacial microsomia, the term used to characterise the typical facial feature of GS patients, as well as the other anomalies associated with this disease, are most likely the result of first and second brachial

arch development problems. These developmental abnormalities appear to have a variety of causes [24, 25].

However, the absence of evident features or the unknowledge of GS characteristics makes the diagnosis difficult and late. The classical features of GS patients involve ocular anomalies, including microphthalmia, anophthalmia, epibulbar dermoid (or lipodermoid) tumours, and eyelid colobomas, aural defects, such as preauricular tags, anotia, microtia and hearing loss, vertebral abnormalities, such as scoliosis, hemivertebrae and cervical fusion, and mandibular hypoplasia 8–12. Facial involvement is usually unilateral, resulting in a marked asymmetry. In the series presented here, the patients were affected unilaterally, and all showed mandibular hypoplasia and vertebral anomaly[26].

Syndromes derived from aberrations in the development of the first and second branchial arches are in the spectrum of GS, including Treacher-Collins syndrome (TCS). The presence of facial asymmetry and far less hypoplasia of the malar bones in GS are the important features to differentiate it from TCS. The TCS-affected patients presented downward slating palpebral fissures, colobomas, zygomatic and mandibular hypoplasia, partial absence of the lower eyelid cilia, and abnormalities of the ears (**Figure 15**).

5.6 Van der Woude syndrome

The disease has an impact on how the face develops. A cleft lip, a cleft palate (an opening in the roof of the mouth) or both are common birth defects in those who have this condition. Affected people frequently have depressions (pits) close to the centre of the lower lip, which may appear moist since salivary and mucous glands are

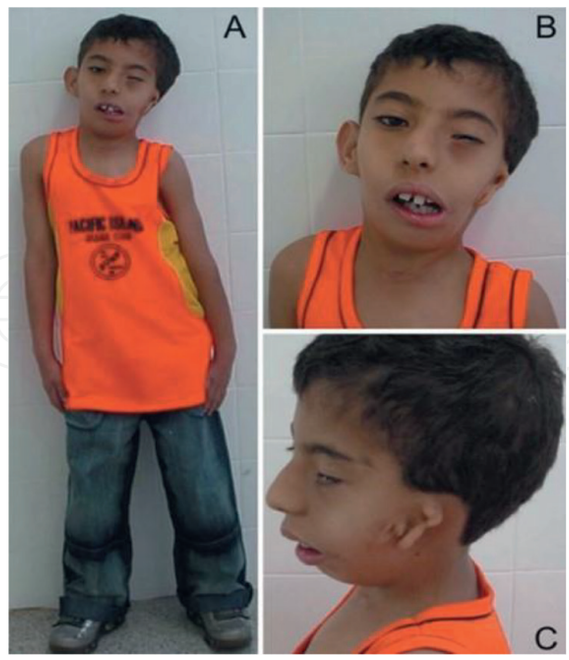

Figure 15.
Skeletal and facial phenotypes of patient #2 of this study. (A) Anterior standing photograph of the patient, demonstrating severe lordoscoliosis. (B) Frontal view of the face showing marked facial asymmetry, malocclusion and eye involvement characterised by microphthalmia and eyelid coloboma of the left eye. (C) Lateral view of the face showing ear malformations, including microtia and preauricular tags (the parents signed informed consent authorising the publication of these pictures).

located there. Also possible are little tissue lumps on the lower lip. Van der Woude syndrome patients can have tooth loss [27].

Like other people with these facial disorders, those who have cleft lip and/or palate also have a greater risk of delayed language development, learning impairments or other modest cognitive issues [28]. The average IQ of individuals with van der Woude syndrome is not significantly different from that of the general population.

5.7 van der Woude syndrome (VWS)

Lip pits* in combination with wiIRF6-related illnesses often fall on a spectrum, ranging from popliteal pterygium syndrome (PPS) at the more severe end to isolated cleft lip and palate and Van der Woude syndrome (VWS) at the moderate end. Rarely, IRF6 pathogenic mutations have also been identified in people with spina bifida (2/192) and nonsyndromic orofacial clefts (18/3811; 0.47 percent). People who have VWS exhibit one or more of the oddities listed below: congenital paramedian lower-lip fistulae (pits), which are typically bilateral, or occasionally tiny mounds with a sinus tract emerging from a mucous gland of the lip, uneven lip (CL) and missing palate (CP). It should be noted that cleft lip with or without cleft palate (CLP) is seen almost twice as frequently as CP alone. Submucous palate cleft (SMCP) has one of the following:

cleft lip with or without cleft palate (CL ± P)
cleft palate (CP)
submucous cleft palate (SMCP)
lip pits* alone and a first-degree relative with CL ± P, CP, or SMCP
CL ± P, CP or SMCP and a first-degree relative with lip pits*
CL or CL + P and CP in the same family
* Lip pits are most often paramedian on the lower lip and can include mounds with a sinus tract leading from a mucous gland of the lip.

5.8 Popliteal pterygium syndrome (PPS)

This syndrome includes the following condition:
popliteal pterygia,
syndactyly,
abnormal external genitalia,
ankyloblepharon,
pyramidal skin on the hallux,
a spectrum of intraoral adhesions, the most severe of which is complete syngnathia.

Musculoskeletal anomalies are rarely reported (e.g. talipes equinovarus, digital reduction, spina bifida occulta, bifid ribs, short sternum).

IRF6-related neural tube defect. Two individuals with an IRF6 pathogenic variant and spina bifida have been reported. Neural tube defects due to an IRF6 pathogenic variant cannot be clinically distinguished from the neural tube defects of other aetiologies. Orofacial cleft was brought on by IRF6. There have been reports of 18 people who had either an orofacial cleft or an IRF6 pathogenic mutation. Clinically, orofacial clefts caused by IRF6 pathogenic variants cannot be discriminated from those caused by other causes.

A proband is diagnosed with an IRF6-related illness based on suggestive evidence, and molecular genetic testing identifies a heterozygous pathogenic mutation in IRF6 [28].

A word is frequently used in clinical genetics to refer to the various methods utilised to pinpoint the molecular causes of genetic illness. Examples of molecular genetic tests include genotyping to identify particular pathogenic variants, gene sequencing to identify pathogenic variations, and amplification or hybridisation techniques to identify copy number variants affecting one or more genes (e.g. qPCR, array CGH, MLPA). Epigenetic alterations are detected using methylation-specific methods.

5.9 Apert syndrome

A hereditary condition known as Apert syndrome is characterised by skeletal deformities. The premature closing of the skull's bones is a major aspect of Apert syndrome (craniosynostosis). Early fusion alters the contour of the head and face and stops the skull from developing normally. The number of fingers and toes that are fused together (syndactyly) varies as well [20].

Many of the distinctive facial characteristics of Apert syndrome are caused by craniosynostosis. Midface hypoplasia, a beaked nose, a wrinkled forehead and a hole in the roof of the mouth are all results of premature skull bone fusion, which prevents the head from growing normally (a cleft palate). Dental problems can result from an underdeveloped upper jaw in people with Apert syndrome.

Many people with Apert syndrome experience vision issues as a result of eye abnormalities, which can include bulging eyes (exophthalmos), wide-set eyes (hypertelorism), outward-facing eyes (downslanting palpebral fissures), eyes that do not look in the same direction (strabismus) and shallow eye sockets (ocular proptosis). Apert syndrome patients with deformed ear structures may experience hearing loss or recurrent ear infections.

People with Apert syndrome who have abnormal facial and cranial structure may also experience breathing issues due to partial airway obstruction. The brain's development is also impacted by craniosynostosis, which may impair intellectual growth. Cognitive abilities range from normal to mild-to-moderate intellectual disability in those with Apert syndrome [29].

Apert syndrome patients exhibit syndactyly of the fingers and toes. Although the severity of the fusion varies, the hands are typically worse off than the feet. Three fingers on each hand and foot are most frequently fused together. The fingers and toes are merged in the most extreme cases. Apert syndrome patients very rarely have extra fingers or toes (polydactyly). Some Apert syndrome sufferers have anomalies in their shoulder or elbow bones. These bone issues might make it difficult to walk around and interfere with daily tasks. While some persons only experience anomalies on one side of the body, others experience abnormalities on both sides.

Hyperhidrosis, oily skin and additional signs and symptoms of Apert syndrome can be present (**Figure 16**).

5.10 Orofacial digital syndrome

The oral-facial-digital syndrome has been linked to the OFD1 gene. Oral-facial-digital syndrome type I is caused by mutations in this gene. Affected family members with a type VII condition were also found to have mutations in the OFD1 gene; nevertheless, experts currently think that type VII and type I disorders are identical [30].

The development of the oral cavity (the mouth and teeth), facial features and digits are all impacted by the oral-facial-digital syndrome, a collection of connected diseases (fingers and toes).

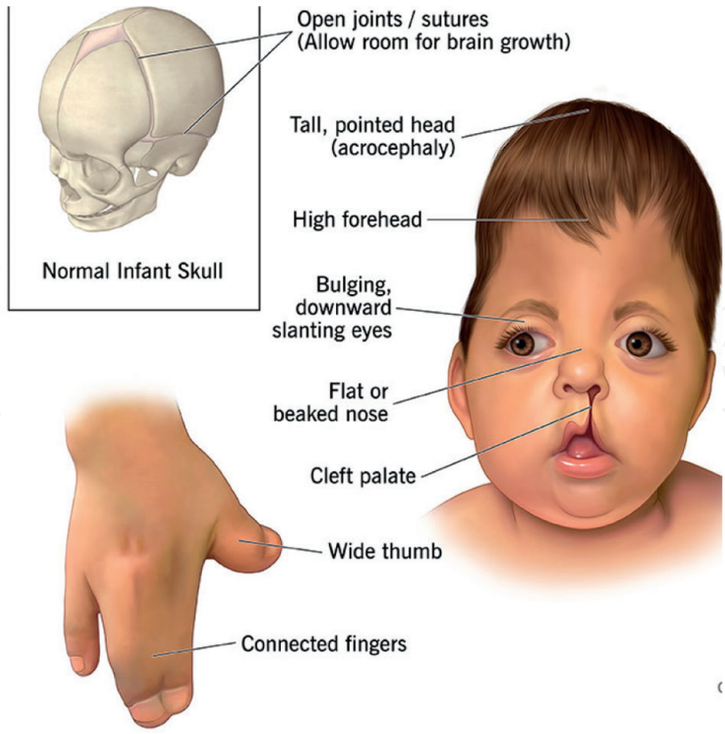

Figure 16.
Salient features of Apert syndrome.

There are at least 13 different possible types of oral-facial-digital syndrome, according to researchers. By their patterns of symptoms and indications, the various categories are categorised. However, there are a lot of overlaps in the characteristics of the different categories, and some types are not clearly defined. The signs and symptoms of oral-facial-digital syndrome vary widely. However, most forms of this disorder involve problems with the development of the oral cavity, facial features and digits [31]. Most forms are also associated with brain abnormalities and some degree of intellectual disability.

A split (cleft) in the tongue, a tongue with an odd-lobed form and the development of noncancerous tumours or nodules on the tongue are among the abnormalities of the oral cavity that occurs in many types of oral-facial-digital syndrome. Additionally, those who are affected might have additional, missing or broken teeth. An aperture in the roof of the mouth is another typical characteristic (a cleft palate). The lip may be abnormally attached to the gums in certain persons with oral-facial-digital syndrome due to bands of excess tissue known as hyperplastic frenula.

Cleft lips, large noses with flat nasal bridges and widely separated eyes are distinctive facial characteristics that are frequently linked to oral-facial-digital syndrome (hypertelorism).

Abnormalities of the digits can affect both the fingers and the toes in people with oral-facial-digital syndrome. These abnormalities include fusion of certain fingers or toes (syndactyly), digits that are shorter than usual (brachydactyly) or digits that are unusually curved (clinodactyly). The presence of extra digits (polydactyly) is also seen in most forms of oral-facial-digital syndrome [31].

Other features occur in only one or a few types of oral-facial digital syndrome. These features help distinguish the different forms of the disorder. For example, the most common form of oral-facial-digital syndrome, type I, is associated with polycystic kidney disease. This kidney disease is characterised by the growth of fluid-filled sacs (cysts) that interfere with the kidneys' ability to filter waste products from the

blood. Other forms of oral-facial-digital syndrome are characterised by neurological problems, particular changes in the structure of the brain, bone abnormalities, vision loss and heart defects.

5.11 Treacher Collins syndrome

SYNONYMUS.
Franceschetti-Zwalen-Klein syndrome.
mandibulofacial dysostosis.
Treacher Collins-Franceschetti syndrome.

Treacher Collins syndrome (TCS), a rare genetic condition, is distinguished by recognisable deformities of the head and face. The zygomatic complex, cheekbones, jaws, palate and mouth are typically underdeveloped in cranial anomalies, which can cause breathing and feeding issues [32]

TCS is mainly brought on by modifications (mutations) in the TCOF1 gene, but it is also linked to changes in the POLR1B, POLR1C or POLR1D genes. While autosomal dominant inheritance is the situation for TCOF1 and POLR1B, autosomal recessive inheritance is the case for POLR1C. On the other hand, POLR1D mutations that are both autosomal dominant and recessive have been linked to TCS. The major characteristic features of TCS encompass certain bones of the face, ears and soft tissues around the eyes. Affected individuals present with distinctive facial features and potentially develop hearing and vision problems. The abnormalities of TCS are typically symmetric (almost identical on both sides of the face) and are present at birth (congenital). Speech and language development can be compromised by hearing loss, cleft palate or jaw and airway problems.

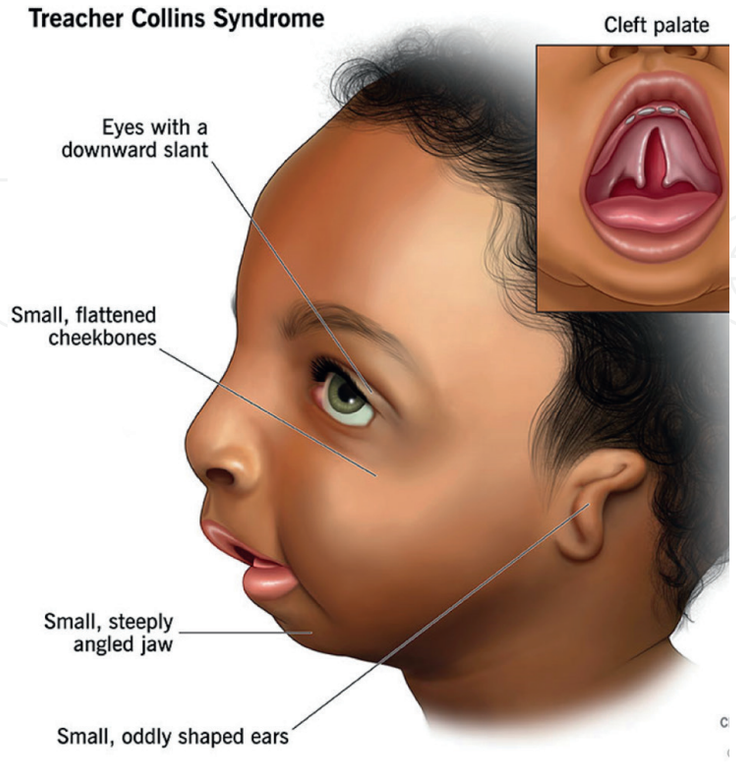

Figure 17.
Salient features of Treacher Collins syndrome.

In children with TCS, the cheekbones are hypoplastic or missing, giving the area of the face a sunken or flat appearance. Due to inadequate development of the lower jaw's (mandible) bone (mandibular hypoplasia), the chin and lower jaw appear abnormally tiny (micrognathia). Obstructive sleep apnoea, which is characterised by frequent, brief pauses in breathing and air movement while sleeping, can affect children. Additionally, dental anomalies such as undeveloped (hypoplastic) or misplaced teeth may result from mouth and jaw deformities (malocclusion). There have also been reports of other dental anomalies, such as tooth agenesis (the absence of teeth), enamel opacity (the clouding or darkening of teeth's enamel) and inappropriate (ectopic) eruption of some upper teeth (maxillary molars) (**Figure 17**) [32, 33].

6. Conclusion

Anomalies of head and neck may arise sporadically or with a strong hereditary predilection. Isolated single anomalies may at times go undiagnosed unless they cause considerable aesthetic and functional concerns. Syndromes usually present more pronounced anomalies; however, treatment to such conditions remain questionable. Systemic involvement may further complicate the management in such cases.

Anomalies that affect dento-facial region are numerous. A thorough knowledge of these anomalies is of great importance to a clinician. Also, a vast amount of cultural, social and personal beliefs regarding the aetiology of such facial deformities and the affected person poses great challenge in providing treatment to such cases.

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