REVIEW OF LITERATURE

1. **Definition:**

Craniofacial anomalies (CFA) are a highly diverse group of complex congenital anomalies (WHO, 2002).

The term craniofacial anomalies literally encompasses all congenital deformities of the cranium and face. More specifically, however, the term has come to imply congenital deformities of the head that interfere with physical and mental well-being (Marsh and Vannier, 1985).

Classification of Craniofacial anomalies

Pathogenic Classification:

I. **Lip and palate:**

A. **International Classification:**

   Classification of the lip, alveolus and palate

   Group 1: Clefts of the anterior palate

      a. Lip: right and/or left
      b. Alveolus: right and/or left

   Group 2: Clefts of the anterior and posterior palates:

      a. Lip: right and/left
      b. Alveolus: right and/or left
      c. Hard palate: right and/or left

   Group 3: Cleft of the posterior palate

      a. Hard palate: right and/palate
      b. Soft palate: median

Rare Facial Clefts

a. Median clefts of the upper lip with or without hypoplasia or aplasia of premaxilla

b. Oblique clefts (oro-orbital)

c. Transverse clefts (oro-auricular)
d. Clefts of the lower lip, nose and other very rare clefts.

**Veau Classification**

Group I (A)
- Defects of the soft palate alone

Group II (B)
- Defects involving the hard and soft palates (not extending anterior to the incisive foramen)

Group III (C)
- Defects involving the palate through to the alveolus

Group IV (D)
- Complete bilateral clefts.

**Kernahan and Stark Classification**

Embryology-based classification system proposed in 1958 that designates the incisive foramen as the dividing line between the primary and secondary palates.

The incisive foramen is a funnel-shaped opening through which neurovascular bundles pass. It is located in the hard palate behind the middle upper teeth (incisors). This structure is an important embryological landmark, which is used to define the boundary between the primary and secondary palate.

- Primary palate includes those structures anterior to the incisive foramen (lip, pre-maxilla, anterior septum).
- Secondary palate includes those structures posterior to the incisive foramen (lateral palatine shelves, soft palate, and uvula).

**Kernahan Classification**

Classification system based on the resemblance of an intra-oral view of a cleft lip and palate to the letter 'Y', proposed in 1971.

The area affected by the cleft is marked on the 'Y' and labelled from 1 to 9, each of which represents a different anatomical structure. Combinations of the numeric values represent the appearance of the cleft lip, alveolus, or palate.
- Areas 1 and 4 represent the right and left side of the nasal floor, respectively.
- Areas 2 and 5 represent the right and left side of the lip, respectively.
- Areas 3 and 6 represent the right and left side of the paired alveolar segment, respectively.
- Area 7 represents the primary palate.
- Areas 8 and 9 represent the secondary palate.

**Harkins' Classification**

1. Cleft of primary palate
   - Cleft lip
   - Alveolar cleft
2. Cleft of secondary palate
   - Soft palate
   - Hard palate
3. Mandibular process clefts
4. Naso-ocular clefts: involving the nose towards the medial canthal region
5. Oro-ocular clefts: extending from the oral commissure towards the palpebral fissure
6. Oro-aural clefts: extending from the oral commissure towards the auricle.

**Spina Classification**

1. Pre-incisive foramen clefts (lip ± alveolus)
   - Unilateral
   - Bilateral
   - Median
2. Trans-incisive foramen cleft (lip, alveolus, palate)
   - Unilateral
   - Bilateral
3. Post-incisive foramen clefts (secondary cleft palate)
4. Atypical (rare) facial clefts.
**Tessier's Classification**

Oro-facial clefts can manifest as:

- Unilateral or bilateral
- Complete, incomplete, or microform (e.g., sub-mucous cleft palate)
- Clefting of the lip with or without the palate, or of the palate in isolation
- Atypical cranio-facial clefts.

A Modification of tessier’s Cleft Classification system

**A. Basic Consideration**

1. The point of reference is the orbit with the clefts found in two different hemispheres.
   a. Those of lower lid are classified as facial clefts
   b. Those of the upper lid are classified as cranial clefts
   c. Combined or craniofacial clefts may occur
2. This system describes both the surface and underlying bony anatomy.
3. The extent of involvement of soft and bony tissue is variable.

**B. The classification**

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II. Face And Skull

The most extensive overall compilation of facial malformation with regards to pathophysiology is that proposed by pruzansky:

1. Sociogenic and iatrogenic factors
2. Intrauterine mechanical factors
3. Focal necrosis
4. Morphokinetic arrest
5. Primary malformations of the skull with secondary effects on the brain
6. Primary malformations of the CNS with secondary deformities of the skull.
7. Chromosomal abnormalities
8. Environmental factors
9. Inborn errors of metabolism
10. Syndromes of unknown etiology

In studying median facial anomalies, the author classifies five types of tissue deficiency disorders which he terms holoprosencephaly:

1. Cyclopi
2. Enthomocephaly
3. Cephalocephaly
4. With median cleft lip (pre maxillary agenesis)
5. With median philtrum- premaxilla anlage

When there is a normal or an excess amount of tissue in the midline structure, DeMeyer favors the term median cleft face syndrome and describes seven features of this entity.

1. Orbital hypertelorism
2. U-shaped frontal hairline
3. Cranium bifid occultum
4. Median cleft of upper lip
5. Median cleft of premaxilla
6. Median cleft of palate
7. Primary telecanthus
A Proposed New classification System

Based on extensive recent experience in the treatment of craniofacial anomalies, anew, practical, and simple classification system is suggested here.

All craniofacial anomalies may be classified into five categories based on etiology, anatomy, and current treatment principles:

I. Clefts
   Centric
   Acentric

II. Synostoses
   Symmetric
   Asymmetric

III. Atrophy- hypoplasia

IV. Neoplasia- hyperplasia

V. Unclassified

All may vary in their manifestation from subtle to extreme.

Unclassified

A. Multiple- Organ Involvement

B. Single- Organ Involvement
   a. Tongue
   b. Nose
   c. Eye and orbit
   d. Lips
   e. Ears
   f. Jaws

Multiple –organ Involvement: These are numerous isolated occurrences which cannot be systematically classified except as described.

Single- organ Involvement

a. Tongue
Macroglossia
Cleft, lobed, fissured

b. Nose
   Transverse groove
   Bifid nose
   Anterior atresia
   Posterior atresia

c. Eyes and orbit
   Epicanthus palpebrisalisisversus
   Simple epicanthus
   Filiform fusion of the eyelids

d. Lips
   Paramedial lip pits or wounds

e. Ear
   Anotia
   Microtia
   Atresia of the external auditory meatus and middle ear abnormalities.
   (Whitaker et al, 1981, Mount 2007)

Classification of Craniosynostosis

Primary
Simple
Nonsyndromic: sagittal, coronal, metopic, lambdoid

Compound
Nonsyndromic: bicoronal
Syndromic: Crouzon’s disease, Apert’s syndrome, Pfeiffer’s disease,
            Saethre-Chotzen syndrome

Secondary
• Metabolic disorders (e.g., hyperthyroidism)
- Malformations (e.g., holoprosencephaly, microcephaly, shunted hydrocephalus, encephalocele)
- Exposure of fetus (e.g., valproic acid, phenytoin)
- Mucopolysaccharidosis (e.g., Hurler’s syndrome, Morquio’s syndrome) (Kabbani et al, 2004)

Etiology of Craniofacial Anomalies:
The possibility of identifying distinct genetic aberrations in craniofacial dysmorphology has improved significantly in recent times. Specific genetic abnormalities resulting in syndromic craniosynostoses and facial dysostoses will be presented later in this issue. In addition to the genetic component, distinct environmental causes have been identified, including infection, radiation, maternal factors, and chemical exposures.

Radiation. High radiation exposure is linked with microcephaly.

Infection. Infection with cytomegalovirus, toxoplasmosis and rubella during pregnancy has been associated with increased chances of facial clefts in newborns as well as concomitant hand and ocular abnormalities.

Maternal idiosyncrasies. Phenylketonuria disorders are more common in females who gave birth to babies with cleft lip/palate. The oculo-auriculo-vertebral (OAV) disorders are more common in offspring borne to mothers having diabetes mellitus. Several studies have linked various factors like maternal weight, general health and age with increased incidence of malformations.

Chemicals. Vitamin deficiency conditions are linked with an increased chances of cleft lip and palate; which may be decreased with vitamin supplementation diets.

Vitamin A and its related compounds such as isotretinoin are thought to be associated with clefts and hemifacial microsomia. Mothers with Vitamin deficiencies are also linked to increase incidence of cleft lip/palate. Maternal smoking is associated with craniosynostosis and facial clefts.
Additional substances are implicated in increased risk of craniofacial anomalies, such as chlorpheniramine, chlordiazepoxide, and nitrofurantoin exposure and Craniosynostosis. (Mount 2007)

**Etiology of Cleft Lip and Palate:**
The etiology of cleft lip/palate is multifactorial and complex in nature. These factors can be classified as genetic factors or environmental factors. (Singh et al, 2011)

**Genetic factors:**
Any disturbances in the functioning of genes those are important in the embryonic craniofacial development can lead to defects in craniofacial development such as cleft lip/palate. A large number of genetic variations have been implicated as causative factor. Mutation or polymorphisms with MSX1, TGFB1, TGFB3, TGFA, RARA, MTHFR, BCLX3, PAX9, FGFR1, FGFR2, TCOF1 etc are shown to be linked with the increased incidence of cleft lip/palate in various populations. Various studies have shown chromosomal aberration in patients with cleft lip/palate. Although family history is important but it is not necessary that the offsprings of cleft parents will develop them. (Singh et al, 2011, 2012)

**Environmental Risk Factors**
A large number of external factors have shown to be associated with increased risk of cleft lip/palate and are related to nutritional factors, illness during pregnancy, habits and teratogenic agents. Some of the important factors are listed below:

**Folic acid deficiency during pregnancy**
Decreased Folic acid intake during pregnancy is associated with increased risk of birth defects and supplementation of folic acid during pregnancy has been suggested. A study has shown that no supplementation of folic acid can lead to a fourfold increase in the risk of birth defects.

**Smoking**
Maternal smoking is thought be associated with increased risk of cleft lip/palate. It is thought to be one of the teratogenic factors.
Alcohol Consumption
Alcohol intake during pregnancy is also a teratogenic factor and is associated with the development of craniofacial anomalies. It is independently associated with increased risk of cleft lip/palate and craniofacial anomalies. Fetal alcohol syndrome is one of the condition that develops due to heavy alcohol consumption during pregnancy.

Obesity and Nutrition
Maternal obesity and poor nutrition are important risk factors for cleft lip and palate.

Medications during Pregnancy
Various drugs have been implicated as teratogenic agents and if taken during pregnancy can lead to various craniofacial defects including cleft lip/palate. Ex- Methotrexate, Isotretinoin and Aspirin to name a few.
(Wong & Hagg, 2004; Murray, 2002; Wehby et al, 2012)

Etiology of Craniosynostosis
Due to recent advances in techniques of genetic analysis, our knowledge of various complex craniofacial anomalies like Craniosynostosis has reached different levels and the genetic component and models to explain the etiology are being proposed. The use of animal models (Knock out mice models) have helped us to study the role of genetics and genetic interactions in the etiology of these complex disorders. Various embryonic origins, perisutural tissue interactions and their receptors, signaling pathways and transcription factors play a vital role in keeping a balance between the proliferation and differentiation of cells in the sutural complex.
(Slater et al, 2008).

Biochemical Factors:
In a study to investigate the role of Indian Hedgehog (Ihh), bone morphogenetic protein (BMP) and Noggin in Craniosynostosis caused by fetal constraint. Expression of BMP-4, Noggin, Histone H4C, Ihh, Sonic Hedgehog (Shh), and Patched 1 (Ptch1) was checked in contrained and control calvarias in animal model. The results suggested that there was decreased expression of Indian Hedgehog (Ihh) and Noggin in constrained induced suture fusion. Further Indian
Hedgehog (Ihh) was shown to regulate Patched 1 (Ptch1) expression during development of cranial sutures and also regulation of osteogenic cell proliferation. (Jacob et al, 2007)

**Environmental Factors**

Current studies point out the role of Nitroso compounds as teratogens and mutagens in animal models. Further it has been suggested that amine drugs with endogenous and exogenous nitrosation reactions can form nitrosocompounds. They can cause various birth defects and congenital anomalies including craniofacial anomalies- cleft lip and or palate and Craniosynostosis.

A study was done to evaluate the outcomes for mothers who were on a drug which undergoes nitrosation against controls who did not receive the drug. The results showed that there were no significant differences between both the groups with regard to fetal death or low birth weight but the risk of major malformations was high in mothers who had drug exposure as compared to controls. (Olshan et al, 1989)

Antiepiletic drugs were associated significantly with increased incidence of congenital malformations especially in the first trimester as compared to the controls. (Jentink et al, 2010).

**Hormonal factors**

In case of juvenile thyrotoxicosis, Craniosynostosis is a common finding. Long term evaluation of the effects of juvenile thyrotoxicosis on central nervous system should be done. Bone is thought to mature early due to increased hormonal level of throid. (Johnsonbaugh et al, 1978)

**Genetic factors**

Syndromic Craniosynostosis shows diverse clinical features. Etiology in terms of genetic causes is also diverse, various genes like fibroblast growth factor receptor genes: FGFR2, FGFR3, TWIST and EFNB1 have been implicated in the etiology of syndromic craniosynostosis. (Jezela-Stanek & Krajewska-Walasek, 2012)
Syndromic craniosynostosis is primarily attributed to genetic basis. There is a strong evidence of involvement of fibroblast growth factor receptor genes: FGFR1, FGFR2, FGFR3 and TWIST genes. These genetic pathways and interactions along with signaling determines the diverse phenotype seen in such cases. (Passos-Bueno, 2008; Lenton, 2005; Wilkie, 1997).

The role of Fibroblast growth factor and fibroblast growth factor receptors is very important in cranial suture growth during embryonic life. There is a proven interaction between FGF, FGFR and TWIST thereby regulating fetal sutural and bone growth. Further FGFR3 mutations are seen in many cases of non syndromic Craniosynostosis. (Moloney t.al, 1997).

**Apert syndrome**
Craniosynostosis is one of the most common congenital conditions. Syndromic varities are mostly caused by mutation in various genes. Genes like FGFR1, FGFR2, FGFR3 and TWIST play a important role. Various animal models are helpful in studying the genetic effects. In this study authors demonstrated role of FGFR2 in Aperts syndrome using mice model. They showed that mutations of neural crest cells is not essential for Craniosynostosis whereas that of mesoderm alone can cause Craniosynostosis. (Holmes & Basilico, 2012).

**Crouzon’s Syndrome:**
The cause of Crouzon syndrome is known to be mutations in the fibroblast growth factor receptor 2 (FGFR2) gene. This gene is also associated with the development of Aperts syndrome and Pfeiffers syndrome. Numerous mutations have been described in the fibroblast growth factor receptor 2 (FGFR2) gene for Crouzons syndrome. (Lin et al, 2012)

**Cleidocranial Dysplasia**
The RUNX2 gene is a physiological regulatory gene implicated in the development of cleidocranial dysplasia (CCD). (Callea et al, 2012).
MSX2 has been implicated in the development of Cleidocranial dysplasia (CCD) (Ott et al, 2012)
Pierre Robin Sequence Complex
Various studies point out the defect to loci 2q24.1-33.3, 4q32-qter, 11q21-23.1, and 17q21-24.3. No significant relation was evident between a particular gene and disease but GAD67, PVRL1 and SOX9 genes are considered to be important in the etiology.

Future studies involving cytogenetics and mutation analyses of suggested genes in nonsyndromic PRS are needed. Efforts should focus on the genomic regions and genes mentioned above. Furthermore, mutation analyses of the genes in populations with nonsyndromic CL/P may provide us with information on the genetic contribution of these genes in the general CL/P population. (Jacobson et al, 2006)

Etiology of Ectodermal Dysplasia
Ectodermal dysplasias appear to represent a primarily genetic disorder whereas only a few genes have been identified as causative agents. Most studies suggested the location at Xq12-q13.1 (XLHED-gene). The development of gene cloning has led to better understanding of genetic disease. The discovery of TNF Transmembrane protein and receptor has shown light on interaction and signaling pathways. Such developments will help in developing method for futuristic possible therapeutic interventions. (Mortier K, 2004)

Treacher’s Collins syndrome:
First and second branchial arches produces important structures in the craniofacial region. Neural crest cell migration plays an important role in development of craniofacial region and defects in neural crest cell migration have been linked to craniofacial anomalies. Various studies employing animal models have shown that maternal exposure to 13-Cisretinoic acid can lead to development of Treachers-collins syndrome. This may be linked to disturbances due to Cis-retenoic acid which induces neural crest migration defects and damage to DNA. TCOF1 has been implicated as the gene responsible for the development of Treachers-collins syndrome. However the exact genetic interactions are not known, but the gene has been mapped at 5q31-34. (Dixon, 1995)
**Hemifacial Microsomia**

The cause is complicated. There are both teratogenic and genetic factors that are implicated. Various agents like thalidomide, retinoic acid and primidone have been documented to be the causative factors. Genetic aberrations induce defects in karyotype. Both autosomal dominant and autosomal recessive inheritance have been thought to be implicated in the disease. (Wang and Andres, 1999)

**Pfeiffer Syndrome**

Pfeiffer syndrome is caused by a defect in FGFR gene. There are two phenotypic variants. Type 1 Pfeiffer Syndrome: It is caused by muattaion in FGFR1 or FGFR2 and represents a milder phenotypic variant. Pfeiffer Syndrome Type 2 and Type 3: They are caused by a mutation in FGFR2. Fibroblast growth factor receptor (FGFR) genes play a major role in maturation of bones. Defect causes prolonged signaling resulting in premature fusion of the bones. (Vogels and Fryns, 2006)

**Facial and Dental features**

**Cleft Lip/Palate:**

**Facial features**

The defects usually associated with cleft lip and palate patients are those of growth in all three planes i.e. vertical, sagittal and transverse plane. The most striking feature in a cleft patient is the sagittal deficiency of the mid face leading to a concave facial profile. The mid facial deficiency is progressive and can be observed in 2 early ages. The major morphologic characteristic in complete cleft lip and palate individuals is the result of altered response of skeletal elements to muscular deformation tendencies. Studies comparing the craniofacial features among cleft individuals have demonstrated significant differences in various parameters when compared with that of the non cleft individuals. Significant variations are observed in Dental and Craniofacial form in Untreated Adult UCLP individuals when compared to non cleft individuals. Facial characteristics were significantly different in sagittal plane when compared to that in the transverse plane. Significant differences were also observed in the dental characteristics between the untreated adult UCLP individuals and the non cleft individuals. (Ravi, 2012)
**Dental anomalies:**
The most affected tooth in cleft area was maxillary lateral incisor; morphologic variation of peg shaped lateral incisor was most common in unilateral cleft lip/palate. There was a correlation between frequency of missing maxillary lateral incisor and severity of the cleft. Supernumerary teeth were more common in unilateral cleft lip/palate followed by unilateral cleft lip alone and unilateral cleft palate. The cleft palate group had fewer dental anomalies compared to others. The maxillary second premolars are affected most commonly outside the maxillary incisor area. (Wu et al, 2011)

**Cranial Features of Craniosynostosis**
The most common types of craniosynostosis are (a) metopic synostosis producing a trigonocephaly deformity, (b) sagittal synostosis producing a scaphocephaly deformity, (c) unilateral coronal synostosis resulting in a plagiocephaly deformity, and (d) bilateral coronal synostosis resulting in a brachycephaly deformity. Lambdoid synostosis is a distinctly uncommon condition that also results in a form of plagiocephaly. Positional (deformational) plagiocephaly of the occiput (a noncraniosynostic, self-correcting deformity) is frequently seen and must be distinguished from Craniosynostosis. (Shin, 2007)

**Dental Features:**
lateral palatal swellings, gingival hypertrophy, multiple tooth agenesis, shovel-shaped incisors, high caries prevalence, early tooth loss, difficult oral hygiene control due to hand malformations, reduced maxillary length, ectopic eruption, tongue thrusting, partial tooth agenesis, high-arched palate, tooth crowding. (Mufalo, 2009)

**Psychological Aspects of patients with craniofacial anomalies**
A study investigating the symptomatology of abnormal appearance using written accounts of 54 patients with different facial defects revealed a similar pattern which can be divided into six parts – difficulties with interpersonal relationships and rationalization, defense mechanisms, unavoidable distress activities, downgrading of self concept, induction and development of self consciousness. (Harris, 1982).
In a review of literature about the social adjustment of people affected with craniofacial anomalies utilizing the electronic database using the key word approach. The results showed that current literature shows that facial appearance has a major influence on social environment, interfering on social contact and development of personality. Psychological support is an important factor in the development and rehabilitation of patients with craniofacial anomalies. (De Oliveira Bastos et al, 2008)

A study was conducted to compare the social skills of preschoolers with and without craniofacial anomalies and to determine what factors are associated with level of social skill. Fifty two children and their families participated in the study. Thirty children were with craniofacial anomalies and rests were matched controls. The children were asked to complete self perception and facial appearance tasks and a social skill interview. Parents and teachers also rated their social skills. The results showed that children with craniofacial anomalies were rated lower in attractiveness and friendliness. On other tasks and social skills there were no differences.(Krueckerberg et al, 1993)

In a study undertaken to investigate the influence of a number of factors (type of malformation, sex of ratee, and sex of rater) on attractiveness and impairment ratings in children with non cleft craniofacial anomalies. The participants were adult eight raters volunteering from a university and two hundred and eight patients with craniofacial anomalies (age group 5.2 to 15.6) .The raters were asked to judge the attractiveness and impairment on a five point scale. The results indicated that the type of malformation, sex of rater and sex of ratee seems to influence judgments on attractiveness and impairment. (Okkerse et al, 2001)

In a review of literature on psychological aspects of cleft lip/palate, the authors have described that psychological stigma in the form of low self esteem, difficulties in social interactions is experienced by these patients. The speech related problems also can subject an individual to poor social interaction. The authors also cite studies addressing educational and vocational issues. The majority of the patients report a high level of satisfaction with surgical results. According to the authors the current cleft psychological research suffers from various methodological weaknesses
including study designs, sample size, time period, satisfaction of parents and patients with treatment. (Turner et al, 1998).

A study was done to evaluate the relationship between symmetry, functional impairments and social adaptation in children with craniofacial deformities. Thirty patients with age from six years to sixteen years were assessed using standard psychological assessment tools including human figure drawing, tasks of emotional adjustments and children’s depression inventory.

Child behavior checklists with parent and teachers forms were used. The results indicated that children with symmetric craniofacial deformities showed poor psychosocial adjustment than those with asymmetric problems, differences between the groups with or without functional impairment were not significant. (Padwa et al, 1991)

A Questionnaire based study that was done to evaluate the attitudes and concerns of British patients with cleft lip/palate and their parents and to assess how closely parents appreciate the problems and concerns of their children. Thirty two patients ranging in age from 16 to 25 years and thirty patients completed the questionnaire. The results suggested that overall the patients were satisfied with the treatment and with the overall facial appearance and speech. The majority felt that their school results and ability to make friends had not been affected at all. In regarding to the specialist, patients ranked surgeon first, orthodontist second and the speech therapist the last. The parents were happy with their child’s overall appearance and speech. Half of them felt that their child had been socially or emotionally affected by the cleft. (Noar JE, 1991)

In a study to assess the psychosocial adjustment in adolescents with craniofacial anomalies and the degree of concordance between parents and adolescents views of youth’s adjustment. The study included 64 adolescents and their parents. They were asked to complete the Youth self report and child behavior checklist. The findings suggested that adolescents with craniofacial anomalies may not be at heightened risk for major adjustment problems, although some may experience deficit in social and academic competence. Parents and children usually displayed great congruence in their reports, however parents identified more problems. (Synder et al, 2005)
A study was done to explore the interaction of gender and age on the self concept of children with cleft lip/palate. The participants consisted of 105 children and adolescents with cleft lip/palate, were asked to complete a questionnaire consisting of medical, demographic variables and Piers –Harris children’s self concept scale. Results showed that most children had average or above average self concept scores. Further adolescents girls experienced a more negative self concept in comparison to younger girls and adolescent boys. (Leonard et al, 1991).

In a study done to analyze the effectiveness of a psychological approach based on cognitive – behavioral principle in the treatment of psychosocial difficulties in the children with altered facial appearance. The sample consisted of 29 children who received therapeutic intervention at “outlook” an organization providing psychosocial care for such patients. Psychometric tools included a semi structured interview schedule, visual analog scales, Child behavior checklist scale. The data was recorded at baseline, post intervention and after a 6 months follow up. The intervention included sessions of social skills and problem solving components. The results indicated that there is a reduction in the frequency of teasing and distress both in classrooms and playgrounds. Parents also reported a reduction in the anxiety levels of children. (Maddern et al, 2006).

A study was done to analyze and asses the factors related to social competence in young adolescents with craniofacial disfigurement. The sample consisted of 48 patients who completed the questionnaires including the self perception scale for adolescents, the social support scale for children and social anxiety scale for children. Their parents completed the child behavior checklist. The degree of disfigurement was rated independently objectively by the investigator. The authors suggested a revised model for predicting social competence including self worth, social competence, social anxiety, severity of disfigurement and perceived parental and peer support. The results suggested that psychological assessment and intervention may be of particular value in social competence. (Shute et al, 2007)

A study was undertaken to evaluate the influence of both congenital and acquired facial anomalies on social adjustment in adults to compare the findings with non affected adults. The study population consisted of fifty nine adults having congenital facial anomalies, fifty nine with
traumatic facial disfigurement and 120 normal adults. They were asked to complete demographic information, social avoidance and distress scale, scale for interpersonal behavior, satisfaction with facial appearance using a visual analog scale, objective assessment of severity of facial disfigurement. The results indicated that there is a impact on social functioning of congenital and acquired group and it differed significantly from reference group. (Van den Elzen , et al, 2012).

A study was done to evaluate the level of satisfaction with facial appearance and its determinants in adults with severe congenital and acquired facial disfigurement and to compare the findings with non disfigured adults. The sample consisted of 59 adults with a rare facial cleft, similar number of adults with a traumatically acquired facial anomaly in adulthood and a control group of two hundred and one normal adults. All the subjects completed the demographic, visual analog scale for assessing the satisfaction, body cathexis scale, Rosenberg self esteem scale, fear of negative evaluation scale. The results suggested that there was no difference with respect to satisfaction with facial appearance in both groups. Demographic factors were important in assessment of level of satisfaction. Fear of negative evaluation by others is one most strong determining factor. (Versenal et al, 2010)

In a brief review on psychological issues in cleft lip and cleft palate the authors have discussed about the stigma experienced by these patients due to their facial appearance, speech and language related problems. Surgical corrections are reported to yield good satisfaction on the part of the patients. While high and unrealistic expectations lead to dissatisfaction. The authors also mention the problems with the current cleft psychological research which includes over reliability on self reported data, not using a longitudinal model, additionally evaluation of parents along with parents.( De souse et al, 2009)

A study was done to examine social anxiety and adjustment in Chinese adult patients with orofacial clefts. The study included 85 adult cleft lip /palate subjects and similar number of their unaffected siblings. Further 85 age and gender matched controls were also included. All the patients were asked to complete fear of negative evaluation, Rosenberg self esteem scale, social avoidance and distress scale, interpersonal support evaluation list. The results indicated that
affected adults have more social anxiety than their siblings or controls. They had also lower self esteem and social support than the two groups. (Berk et al, 2001)

A study was performed to assess the satisfaction of patients and their parents with facial appearance and its relation with psychological functioning. The study subjects consisted of 111 subjects and 62 parents. Facial appearance was rated using a subjective ordinal scale while psychological adjustment by childhood experience questionnaire. The results indicated that younger subjects of age 10 years were less satisfied with their facial appearance than 15 years old. Subjects affected by a visible problem are more dissatisfied than those with invisible impairments. Self satisfaction also correlated with psychological functioning in case of 10 and 15 years old. (Thomas et al, 1997)

A study was undertaken in order to develop a new psychometric instrument for the assessment of patients with problems of appearance. A scale was formulated with 136 items that were based on experiences of patients and clinical experience from plastic surgery patients. Then further workup was done with a longitudinal study and the validity and reliability of the scale was confirmed. Further refinement of the items lead to the two versions of the scale- a long and elaborated version with 59 items – the DAS59 and a short version with 24 items – The DAS24. The current study of the author describes that DAS 59 is a very sensitive, specific tool on the other hand it has good validity and reliability to be used for individuals with appearance problems. (Harris, Carr, 2001)

A study was done to produce a questionnaire for knowing the psychosocial impact of dental aesthetics in young adults. The study subjects were 194 young adults who were questioned using a list of 23 items that relate to the subject. Aesthetic component of the index of orthodontic treatment needs was used to rate the dental aesthetics appearance of the subject by the subject itself and interviewer separately. Perception of occlusion scale and a modified version of the dental aesthetic index were also used. The results suggested proposed instrument that meets the criterion of factorial stability across samples and criterion related validity and reliability. (Klages et al, 2006)
A study was conducted to develop a psychometric instrument to assess the patients and their parents’ expectation before orthodontic treatment. The sample consisted of 70 patients and one of their parents. The authors utilized a two step method to identify themes and concepts using open ended interview so as to formulate a experimental version of questionnaire. Finally 10 items, along with sub items were developed that utilized a visual analog scale as a response. The questionnaire had good validity and reliability. (Sayers, Newton, 2006)