# A STUDY OF CASES OF HEREDITORY ECTODERMAL DYSPLASIA -A RARE DENTAL ANOMALY

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**ABSTRACT**

Hereditary ectodermal dysplasia is a group of disorder running in the family where more than one manifestation occurs involving skin, nail, hair, glands and teeth In the present study, five cases were detected in district of Bangalore,Karnataka and studied in detail. Out of them three were girls and two boys showing manifestation dysplasia of teeth, skin & sweat glands between the age groups of 5 years to 14 years of age. There are three girls between 5 to 18 years showing oligodentia(0.13%) in 2 girls and anodentia in one girl(0.67%) associated with periorbital wrinkling and mild mid facial hypoplasia.The other 2 were boys between 8years and 15 years of age showed oligodentia, anhydosis ,brittle nails with vertical ridges, and 15 years old boy also showed periorbital wrinkling. The mothers of these five patients were also studied. Consanguity along with heredity and hypertension has played a vital role in the development of ectodermal dysplasia .These 5 cases were compared and correlated with available literatures.

**KEY WORDS:** Ectodermal dysplasia,-- Oligodontia- Anhydrosis -Consanguineous couples Sparse distribution of hair - xerostomia

## INTRODUCTION:

Hereditary ectodermal dysplasia is a group of inherited disorders primarily involving defects in the development 2 or more tissues derived from the embryonic ectoderm .The tissues primarily involved are skin, hair,nails,eccrine glands & teeth. The disorders include sparse distribution of blonde hair, associated with decreased density of eyebrow, eyelashes There may be fine wrinkling and hyper pigmentation in the perioccular skin. There is hypoplasia 0f the midface ,protuberant face, .There is varying degree of xerostomia. and dystrophic brittle nails. The skin in these patients has lost power of regulation of body temperature Hence they may get fever of unknown aetiology.[1]

AETIOLOGY;-Hereditary ectodermal dysplasia is a syndrome by aberrant development of ectodermal derivatives that occurs during early part intrauterine life..It is because of genes that may undergo mutation or deletion Ectodermal dysplasia are autosomal recessive disorders .The gene that cause hidrotic ectodermal dysplasia is GJB6. GJB6 has been mapped to pericentrometric location of chromosome 13q Genetic mutation in the PVRL1has been found in ectodermal dysplasia giving rise to cleft palate.[2] Linkage studies have confirmed the location of gene at XQ12-Q13-:1(XLHED-gene ).This localization of gene is the more accurate diagnosis of this disorder both in carrier female and in prenatal screening.[3]

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## MATERIALS AND METHODS:

Out of 1500 patients screened for the presence of any dental anomalies, 5 anomalies with ectodermal dysplasia were detected presenting with different features in Bangalore district of Karnataka constituted the materials for the present study . There were three females and two males ,the age group ranging from 5 years to 18 years. The following histories were taken in detail, they are i)History of drug intake during 1st trimester.ii)Family history of consanguity, iii)Family history of hypertension ,diabetes, and seizures.iv)Any history of still birth, repeated abortions ,hydramnios,

All the five cases were examined in detail for the presence of anomalies. Examination of oral cavity, skin, eyelids were carefully done Later they were subjected to relevant investigations like hematological examination, urine analysis, and X-Rays with some special investigations

## ROUTINE INVESTIGATIONS IN MOTHERS:

**A)BLOOD BIOCHEMISTRY** : Test were done-that includes ,fasting blood sugar post prandial,blood urea serum creatinine,serum cholesterol,total

cholesterol,total calcium,lipid profile,liver function test,creatinine phosphokinase,(CPK),alkaline phosphotase,acid phosphotase,amylase total bilirubin total protein,,sodium ,potassium and calcium

**B)HAEMATOLOGY:**

Haemoglobin Percentages (Hb%), RBC- TC) Differential Count (D.C) of leucocytes,Platelet Count - Bleeding Time (BT),Clotting Time (CT)Prothrombin Time (PT)Reticulocyte Count,ABO Grouping Rh type ,Peripheral Smear in study of anemia ,Erythrocyte Sedimentation Rate (E.S.R.) were done

SPECIAL INVESTIGATIONS:- Antenatal Ultrasound was done in all patients (mothers) with karyotyping postnatally

**OBSERVATIONS:**

CASE-1;-A female patient aged 18 years came with history of less number of teeth in the oral cavity Oligodontia with congenital absence of teeth since childhood There was loss of sweating. On examination, she was found to have erupted teeth which were less in number along with congenital absence of teeth.Erupted teeth were peg shaped or screw shaped. There was sparse distribution of hair over the scalp and over the extremities She was normally built and nourished .Nails and salivary glands were normal There was no other associated anomalies -like cleft palate or cleft lip and no impairment in hearing nor in vision but there was periorbital wrinkling of skin . All her fingers and toes were normal. Her intelligence was normal.There was a similar history in their family tree

CASE-2:-12 years old girl came to out patient department with history of non eruption of few teeth along with wide spacing of erupted teeth - oligodentia, Erupted teeth were screw shaped ,associated with on and off fever..She was treated for Pyrexia of unknown origin.but fever did not respond to any treatment .Later she was referred to skin department where skin was taken for histopathological studies..The report of skin biopsy showed reduction in sweat glands hair follicles and sebaceous glands..There was reduced density of eye brows and eye lashes. Her skin was unable to regulate body temperature. hence she was getting fever on and off There was no anomaly of nails, and palate

CASE-3:-A Five years old girl came with history of complete congenital absence of teeth- ANODENTIA.The girl was ,short statured with mild

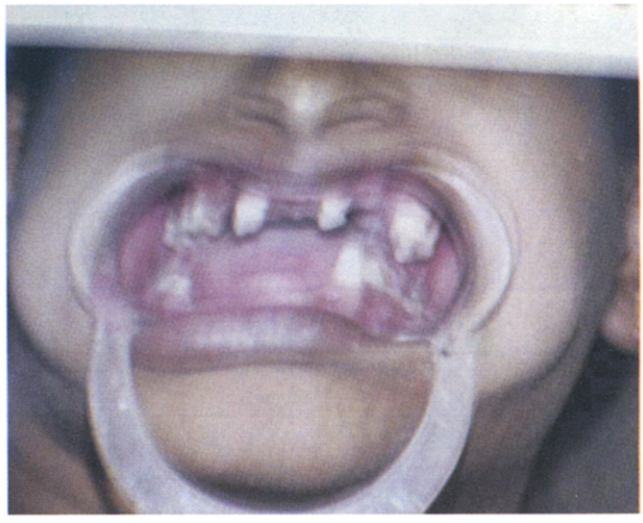

**Fig.1:** Photographs Showing partial absence of teeth since birth Oligodentia

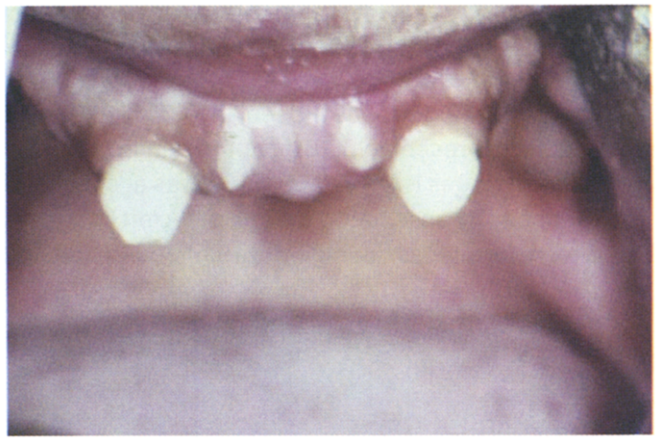

**Fig-2:** Photographs showing oligodentia in 8 years old boy.

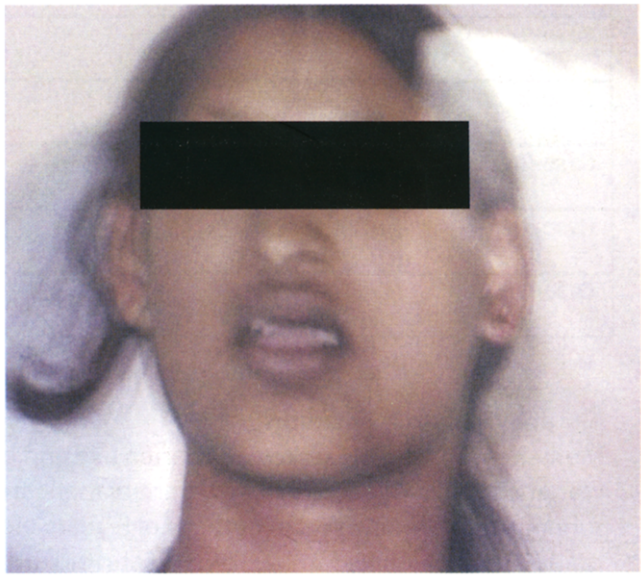

**Fig -3:** Photographs showing congenital absence of teeth in a years old female with long neck & sparse distribution of hairs Eyebrows .

| case s | Oligodentia | Anodentia | Anhydrosis | Periorbital wrinkling of skin | Brittle nails with vertical ridges | mild mid facial hypoplasia, | cleft palate /cleft lip. |
|---|---|---|---|---|---|---|---|
| case -1 | PRESENT | ------- | PRESENT | ---------- | ---------- | ---------- | --------- |
| case -2 | PRESENT | ------- | -------- | ---------- | ---------- | ---------- | --------- |
| case -3 | -------- | PRESENT | ---------- | PRESENT | ---------- | PRESENT | ------- |
| case -4 | PRESENT | --------- | PRESENT | ---------- | PRESENT | ---------- | --------- |
| case -5 | PRESENT | --------- | PRESENT | PRESENT | PRESENT | ---------- | --------- |

**Table-1**,showing clinical features in all 5 cases of Ectodermal Dysplasia

Above tables shows

i)Incidence of Oligodentia seen in 0.24% cases ii).Incidence of Anodentitia is 0.59% , iii)Incidence of Anhydrosis is present in 0.18%, iv)Incidence of Periorbital wrinkling of skin is 0.12%, v)Incidence of Brittle nails with vertical ridges is 0.12%, vi)There was a single case of mild mid facial hypoplasia(0.59%). vii)But there were no incidences of associated cleft palate /cleft lip

There were two cases having brittle nails, seen both in boys of 8 and 15 years old respectively.

| MOTHERS OF DENTAL ANOMALOUS SUBJECTS | Hypertension | diabetes | epilepsy | Heredity | Consanguity |
|---|---|---|---|---|---|
| Case-1 | PRESENT | ---------- | ---------- | ---------- | PRESENT |
| Case-2 | ---------- | PRESENT | ---------- | PRESENT | ---------- |
| Case-3 | ---------- | ---------- | PRESENT | ---------- | PRESENT |
| Case-4 | PRESENT | ---------- | ---------- | ---------- | ---------- |
| Case-5 | ---------- | ---------- | ---------- | PRESENT | PRESENT |

**Table-2**,showing maternal history of 5 cases :-

The above table-2, illustrates

Mother of 18 years old girl was hypertensive and grade 2 consanguity,where as 12 years old girl's mother was known diabetic & she was on ant diabetic treatment and also she gives history of similar anomaly running in the family..While Mother's of 5 years old girl was known epileptic & she was on treatment with grade 2 consanguity.Mother of 8 years old boy was hypertensive. She was s on antihypertensive treatment. Mother of 15 years old boy give history of heredity and consanguity, but degree of consanguity was not clear.

facial hypoplasia a long oval face On clinical examination she was found to have peculiar hyperpigmentation, sparse distribution of hairs in the scalp and reduced density of eye brows and eye lashes,Ultrasound was done which revealed infantile uterus Peri orbital wrinkling of skin was present There was no anomaly of nails, and palate There was no history of fever .Similar history was present in the family in the past

CASE -4;-A 8 year old boy was shown at out patient department with history of eruption of only few teeth-OLIGODENTIA and decreased tears in his eyes. associated with decreased sweating -- ANHYDROSIS. On examination, he was short statured,. Erupted teeth were screw or peg shaped.Nails were brittle There was minimal secretion in the salivary glands .But there was no other anomalies like cleft palate and cleft lip..Fingers, toes ,intelligence hair distribution in the scalp were NORMAL There was no family history of similar complaints.

CASE-5:-A 15 years boy came with h/o oligodentia ,Anhydrosis in frontal region with sparse distribution of hair in the scalp..All the nails were brittle with vertical ridges, unable to cut with nail cutters Periorbital wrinkling of skin was present Oral examination showed only 2 peg shaped teeth He was diagnosed to have ectodermal dysplasia only on clinical findings and histopatholgical report. He was very intelligent and stood 1st in the class. There was family history of similar complaints .All three generations had similar complaints with varied degree of severity

## DISCUSSION

**CLINICAL FEATURES:-** Hereditary ectodermal dysplasia is characterized by three signs Triad of signs namely i)Sparse distribution of hair on body and scalp-known as Atrichosis or hypotrichosis ii)Abnormal or missing of teeth Anodentia or oligodontia iii)Lack of sweating due to absence of sweat glands-Anhidrosis or Hypohidrosis.[4]

Incidence in male is said to be 1 in 100,000 births and carrier incidence is 17.3 in 100,000.women .Most patients have normal expectancy of life and normal intelligence

The most characteristic feature in man is reduced number and abnormal shape of teeth. The delay in eruption of teeth is first diagnosis .Some infants have premature look with scaling of skin which is a clue for diagnosis..The boys have old man facies .There is reduction of sweat glands along with sparse distribution of hair in scalp and body .There is also lack of eye lashes and eye brows.Females are carriers One third carriers are healthy, other third show mild symptoms and another third show full symptoms symptoms.[4]There are clinically and genetically 160 types of hereditary ectodemal dysplasia[5] Hereditary ectodermal dysplasia is a common variety of ectodermal dysplasia(80%) characterized by hypoplasia of skin, teeth, & hair.[6] It is missing teeth or delay in eruption brings worry to parents and this can be diagnosed in second year.[7] Incidence is estimated to be 1 in 10,0000 live births in males[8] The screening limit for eruption of 1st tooth is one year three months or 15 months Hypondontia is said to be very common in affected people ..Sometimes both the jaws may be toothless. Regarding sweating some individuals sweat in certain regions of the body namely palm axilla and sole .In such affected individuals, there is reduced number of sweat glands and there is danger of hyperthermia sometimes associated with sudden death9. In 30% of populations, there is both mortality and morbidity in affected patient. Especially in males too in 1st and 2nd year. due to fever and chest infection[10].Majority of patients complain of dryness of mucous membrane in oral cavity and nasal cavity due decreased secretion of saliva[11]. In few patients of ectodermal dysplasia, there may be craniofacial deformities due to variable amount of missing osseous or dental tissues. Clinically there may be fontal fullness with prominent supraorbital ridge There is also depression of nasal bridge called saddled nose There is also depression and hypoplasia of mid face giving the appearance of Dished In. There may be associated pointed chin , evertion and ,protuberance of lips[12] Cephalometric analysis and anthropometric studies have been performed which revealed reduced facial dimensions decreased facial height and variable pattern in facial width There is retrusion of maxilla than mandible There is also significant reduction in alar nasal width and mouth width[13] Hyperthermia has caused brain damage and mental retatardation .There will be diminution of subcutaneous fat and one third of population will have breast abnormalities.[14] The mapping of gene to x linked gene has given new possibilities of detection of carriers of XLHED by molecular genetics[15]

Hereditory Ectodermal Dysplasia is one of the genetic disorders first described by Thurnam in

184816 ..Later,it was described by Darwin[17] In 1921.Thadani assigned it to X chromosome[18]. In 1929 ,Weech, coined it as Ectodermal Dysplasia[19].

Hereditory Ectodermal Dysplasia was studied in 17 years old boy from Trivandrum born to Non Consanguineous couple.He had congenital absence of teeth On examination of oral cavity there was complete absence of mandibular teeth & anodentia of maxillary arch .Teeth of 13,11,21 of maxillary arch were conical in shape There was history of delayed eruption of teeth. There was also bossing of frontal bone, saddled shaped nose ,& everted lips. There was history of complete absence of sweating since birth On grounds of dental & clinical findings ,he was diagnosed to have Hypohydrotic Ectodermal dysplasia whichis Xlonked disorder[20]

Ectodermal Dysplasia was seen in 32 years old male with history of multiple missing teeth since childhood .He had delayed eruption of deciduous teeth & permanent teeth & loss of sweating ,dry skin associated with intolerance to heat There was prominent supra orbital ridges.His skin was warm & dry .He also had sparse distribution of eye lashes in both upper & lower eye lids There was multiple missing teeth both in mandibular & maxillary arches Orthopantomograph was done which showed multiple missing teeth.He was Diagnosed to have Hereditory Ectodermal Dysplasia[21]

Lu P D & Schaffer.JV reported Hypohidrotic Ectodermal Dysplasia (HED) in 3 children that included 2 sisters & a unrelated boy Each patient presented with hypodroisis, oligodontia, sparse distribution of hair , conical teeth, dermatitis which was eczematous, saddle shaped nose. There was also bossulation of frontal bone & prominent lips .Defects in Ectodysplasin signal transduction pathway result in hypohidrotic Ectodermal Dysplasia (HED) . Mutation of genes encoding the ligand Ectodysplasin A(EDA) under lie classic, X linked recessive HED, while genetic mutation encoding EDA receptors & adaptor protein result in autosomal dominant & autosomal recessive forms of hypohidrotic Ectodermal Dysplasia[22]

There is one form of disorder known as Anhidrotic which presents a classified triad namely hypohidrosis,Hypotrichosis,hypodontia. Hereditory Ectodermal Dysplasia was reported in 11 years old Indian with history of delayed eruption of tooth On examination of oral cavity,there was malformed teeth present since child hood & missing lower teeth .When he was 11days old he had PYREXIA Unknown Origin There was sparse distribution of hair fine scalp hair & he had intolerance to heat There was no history of similar complaints in the family. Other findings were ,sparse eye lashes & eye brows with fine brittle scalp hair associated square fore head frontal bossulation & depressed nasal bridge everted lower lip, dry rough skin ,but his nails of hands & feet were normal[23]

In boy of 12 years old there was history of high grade fever on & off & difficulty in mastication .There was history of parchment like skin at birth .On examination there was sparse distribution of scalp hairs .Eyes brows & eye lashes were absent .There was malar hypoplasia ,nasal bridged was depressed .& in the frontal bone there was bossulation.In the periorbital region there was hypo pigmented skin. Skin was rough with absence of sweating .

In another case ,of 11 years old female ,with abnormalities of teeth ,There was history of fever in infancy & absence of sweating & there was also over heating of skin during summer.Clinical examination of the patient revealed , fine distribution of sparse scalp hair, eyebrows, and eyelashes. The skin was smooth and dry with mild periorbital andperioral hyperpigmentation. Along with bossing of frontal bone , supraorbital ridges were prominent , and with a depressed nasal bridge[24]

Lamartine (2003)stated ectodermal dysplasia as a large & complex disease characterized by defects in hairs nails teeth & also sweat gland ,He pointed out that out of 170 types ,only 30 types have got causative gene that can be explained at molecular level.He tried to classify causative genes into 4 major functional subgroups namely cell cell communication & signaling ,adhesion, transcription regulation & development[25]

Hypohydrotic ectodermal Dysplasia is also known as Christ-Siemens Touraine syndrome It is a rare disorder of genetic origin involving several ectodemal structures ectodermal structures.It is inherited as inherited as X-linked recessive trait,where females are carriers & males are affected.This syndrome has v manifestations varying from individual to individual involving skin, hair, nail, sweat and sebaceous glands. Hypohydrotic Ectodermal Dysplasia with classical features in two siblings is reported here.[26]

8-year-old boy with ectodermal dysplasia complained of difficulty in chewing Oral examination revealed absence of maxillary and mandibular teethHis delivery was normal .There was no family history of absence of teeth .But there was history of

high fever, heat intolerance , and absence of sweating On physical examination ,he was short & underweight, low I.Q,. sparse eyebrows,brittle nails, dry and rough skin.There were also bossing of frontal bone , saddle nose, reduced vertical dimension of face was reduced due to complete absence of teeth .Tongue & palate were normal [27]

**PRESENT STUDY :**

Has shown congenital absence of teeth and presence of peg or screw shaped teeth ranging from 2-6 in number-OLIGODONTIA and there is also history of high grade fever in a 12 years old girl with fever not responding to any treatment but sweating was present in one half of frontal region palms and soles not in axillae.-ANHYDROSIS. Study has shown only morbidity but no mortality .But there was family history of mortality in one of affected baby due to ectodermal dysplasia.. This study has also shown a mid facial deformity in 5 years old girl with a long oval face On clinical examination she was found to have peculiar hyper pigmentation, with sparse distribution of hairs in the scalp and reduced density of eye brows and eye lashes, Periorbital wrinkling of skin Ultrasound was done which revealed infantile uterus with no anomaly of nails, palate Other dimensions of face were within normal limits. All the 5 patients complain of dryness of mucous membrane of oral cavity and nasal cavity due to decreased secretion of saliva..In this study there was no case of brain damage nor is mental retardation There is no incidence of abnormalities of breast. All the levels of calcium, blood urea, uric acid were normal in mothers of anomalous subjects .Only two mothers were suffering from systemic hypertension and not from preeclampsia toxaemia.They are on treatment for systemic hypertension.

**CONCLUSION:**

This study on ectodermal dysplasia gives adequate information regarding morbidity of ectodermal derivatives not only to dental surgeons but also to the Anatomists, dermatologists, paediatitrians.Proper health education should be given to consangunious couples, mothers of ectodermal dysplasia through health education in order to avoid further pregnancies or its impact on minds of consangunious couple. Heredity and consanguity have played prominent role in the development of ectodermal dysplasia.

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