WORKING DIAGNOSIS FOR A CLINICAL SCENARIO -A CHALLENGING ASPECT

[1] Dr. N. Santana MDS,[2] Dr. Kumari. M MDS,[3] Dr. Sangeetha. R MDS,[4] Dr. Sai Charan K.V MDS,[5] Dr.Gayathri. V.S MDS

[1] Professor and HOD,[2] Corresponding author,[3] Professor and guide,[4][5] 2nd year post graduate student

[1][2][3][4][5] Department of oral medicine and radiology, Ragas dental college and hospital..

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ABSTRACT

Stature is the term which represents the quantifiable measure of height which varies widely among the racial group with an equitable normal distribution. Among the various patient encountered by the clinical practitioners, The case of short stature is relatively few; clinicians must be able to distinguish pathologic short stature due to various causes. There are empirically infinite different causes of short stature, and the clinical evaluation of short stature requires an extensive variety of clinical diagnosis, radiographs, pathologic and biochemical assays. Even though definite management to promote growth is offered only in individuals with the endocrine disorders, the acquired nutritional, emotional and chronic disease states, diagnosis of the specific form of short stature might illustrate great significance in being able to prevent complications and this manuscript discusses about the clinical scenario provisionally diagnosed to be achondroplasia.

Key words: short stature, growth, achondroplasia, proportionate , disproportionate.

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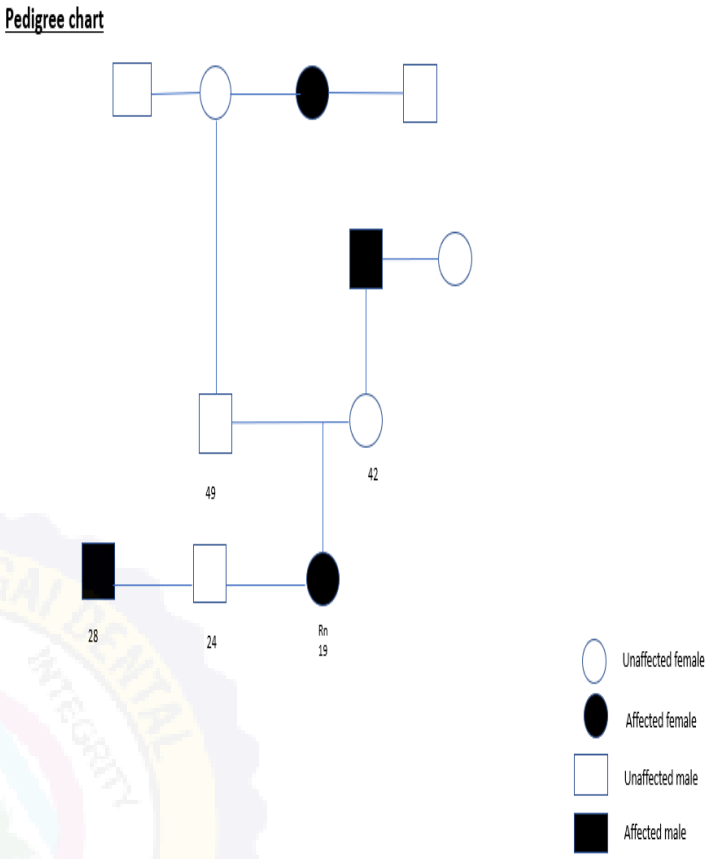
INTRODUCTION

"Short stature is a symbol of disease, disability, and social humiliation causing psychological stress. It is important to diagnose earlier and treat properly. Short stature may result from skeletal dysplasias, endocrine disorders, genetic disorders or would be the result of malnutrition and chronic illnesses. A troop effort of the healthcare professionals like paediatricians, endocrinologists, radiologists, stomatologist and pathologists is deemed necessary to diagnose, treat and monitor numerous critical circumstances related with growth abnormality.

Short stature may be proportionate or disproportionate. Disproportionate dwarfism is characterised by one or many parts of body being comparatively large or small, along with growth deviation in other areas being apparent. This condition is classically caused by one or more genetic disorders involving bone or cartilage development, as seen in skeletal dysplasia's. Hypotonia or otherwise low muscle tone, is common condition where IQ is always normal. In proportionate dwarfism, the body appears normally proportioned, but is bizarrely small. The shortness with proportional body parts along with growth hormone deficiency is called pituitary dwarfism. While in disproportionate dwarfism, intelligence will be spared.This article discuss about the case of short stature which has diagnosed based on clinic examination and radiographic findings in our department.(2)

CASE SCENERIO

A 19 years old female reported to our department with the chief complaint of pain in right lower back tooth region. The patient had short stature and had a waddling gait. The patient gave positive family history of her elder brother and her maternal grandfather and paternal grand aunt affected in a similar manner. Her parents had non-consanguineous marriage, got conceived by natural conception and delivered normally at 9 months of gestation. The Patient was well-oriented, well-nourished and cooperative



General physical examination revealed short stature with shortening of the arms and legs with rhizomelic features on right side and average sized trunk. Lumbar lordosis and short stubby fingers with trident hand configuration were also evident. Anthropometry (at age 19 years) revealed a height of 117cm and weight of 39kg. The head circumferences was 54.5cm, right hand was 54cm, left hand was 46cm and right leg was 66cm, left leg was 69cm and inter canthal distance, was 4.5cm. Her milestones were attained at appropriate age. She was done with proper schooling and graduation .Her IQ was within normal range. Her vital signs were within normal limits. She had delayed onset of puberty with menstrual cycle which is normal in frequency ,flow and rate.

Extra oral examination revealed facial features such as, frontal bossing, flattened nasal bridge, midfacial hypoplasia are seen, neck is normal, chest appears to be pigeon shape , rhizomelic shortening of limbs, average sized trunk and normal abdomen and scoliosis of the spine is evident.

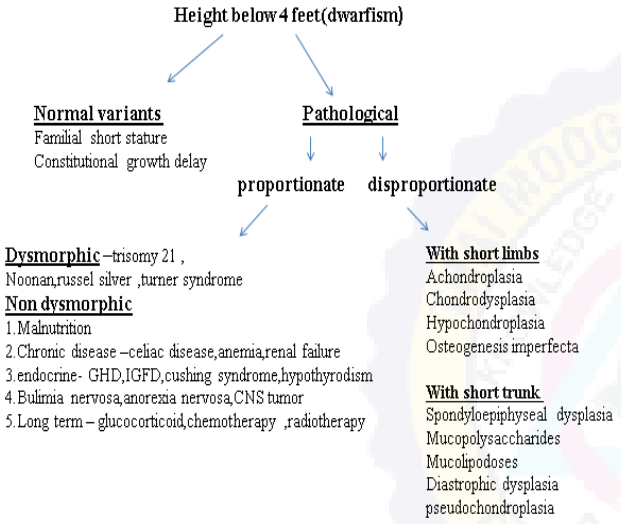
Intraoral examination revealed presence of dental caries 46, interdental spacing, flaring of mandibular anterior tooth and enamel hypoplasia in right upper quadrant

On radiographic examination, her orthopantomogram showed dental caries with periapical periodontitis in relation to 46. The mandible appeared normal. The hand-wrist radiograph showed a trident hand configuration. The PA view of skull revealed an enlarged calvarium and all other structures appeared normal.

Based on the revealed history, clinical presentation ,radiological investigations and external features achondroplasia was considered as a specific diagnosis for the short stature, differential diagnosis of hypochondroplasia, chondrodysplasia, osteogenesis imperfecta was made.

DISCUSSION

DIAGNOSTIC TREE OF SHORT STATURE



During adolescence normal height range should be estimated , which ranges around male (54 to 74 inches) ,female (50 to 68 inches). Where as in our case height is 46 inches. This confirms the patient has short stature. As there are various reason for short stature, conditions like endocrine disorders, syndromes, chronic disease, bone diseases are ruled as the patient do not have any dysmorphic features .Other findings in clinical examination like frontal bossing ,flattened nasal bridge ,midfacial hypoplasia ,rhizomelic shortening of limbs, trident hand ,scoliosis were evident ,which made us to work on differently diagnosis for each finding .

Table 1: comparing the patients clinical presentation with the specific disorder of same presentation

SALIENT FEATURES	DISEASES
Frontal bossing	Osteogenesis imperfecta, Rickets, Acromegaly, Cleidocranial dysplasia, Gardner syndrome, Cogenital syphilis, Crouzon syndrome, Ectodermal dysplasia, Hallerman steriff syndrome β thalesemia
Flatenned nasal bridge	Cleidocranial Dystosis, Downs syndrome, Achondroplasia
Short limbs	Achondroplasia, Hypochondroplasia, Kyphomelic dysplasia
Midfacial hypoplasia	Pierre robin syndrome, Fetal alcohol syndrome Achondroplasia, Crouzon syndrome
Trident hand	Achondroplasia, Anauxetic dysplasia ,Hypochondroplasia

On elaboration the family history the patients elder brother is affected with same condition but other family members were apparently normal .Secondly patient stature was disproportionate .we noticed a disproportionate short stature is characterized by a short limb but an average sized trunk so we exclude the differential diagnosis under proportionate short stature(ref tab 1).on auxiliary working on disproportionate short stature we aimed to look disproportionate with short limb and short trunk , The conditions with short trunk like spondyloepiphyseal dysplasia findings like flat face ,cleft palate, barrel shaped chest, muscle ,hypotonia, senso-neural deafness ,arm is slightly longer than torso, cleft palate ,while hypotonia not evident in above case(6)

In mucopolysaccharides ,general manifestations such as infiltrated face, short stature ,thick eyebrows, joint hypermobility ,skeletal deformities ,cardiac manifestations ,respiratory manifestations ,retardation of psychomotor skills and respiratory illness is present ,in our case no RS,CVS abnormalities are seen, so condition is rejected.(7)

In the condition of Mucolipidosis, along with short stature, myoclonus, cherry macules, puffy eyelids, tremors, seizures, liver and spleen enlargement are the characteristic feature , while these findings are absent in our case the condition is rejected.

In condition of diastrophic dysplasia features like normal size skull ,hitch-hicker thumb, early onset of osteoarthritis ,ulnar deviation of fingers and gap between first and second toes and clubfoot .no finger abnormalities is seen in our case(8)

In Pseudo-chondroplasia, characterized by normal length at birth and normal face, joint pain is appreciated during childhood,no joint pain is noted in our case.

In our case, the individual presented with short limbs with average sized trunk hence the differential diagnosis with short trunk is ruled out.

The conditions with short limbs with disproportionate stature includes achondroplasia, chondrodysplasia, Metaphyseal chondrodysplasia, Osteogenesis imperfecta

In Osteogenesis imperfecta, findings like blue sclera, hearing loss, deformed teeth, stature is normal or mildly reduced, abnormal dentition, CNS & CVS abnormalities are witnessed. hence the particular case the stature is short and don't have any above mentioned features ,OI is excluded.b(3)

The rhizomelic limb shortening features like achondroplasia and pseudochondroplasia. Pseudochondroplasia is ruled out as it has short trunk, our case has average sized trunk(6)

The trident hand configuration is found in achondroplasia, Hypochondroplasia and anauxetic dysplasia, while anauxetic dysplasia is ruled out as features like deformation of the spinal cord and joint deformity is absent (7)

The features like midfacial hypoplasia, frontal bossing, rhizomelic limb shortening, disproportionate dwarfism, lumbar lordosis, trident hand configuration is evident in our case, which are exclusively found in achondroplasia. Hence all the conditions are ruled out. The clinical features mostly mimicks the condition of achondroplasia, the provisional diagnosis of achondroplasia had been decided.(8)

Achondroplasia is reflected as a non-lethal form of chondrodysplasia. It is the notable common form of skeletal dysplasia and is categorized by short limb dwarfism, affecting the growth of the tubular bones, spine and skull. The prevalence of the condition is valued to be 1 in 15,000 of population. It is engrained as autosomal dominant trait with complete penetrance. The common mutation of the FGFR3 gene, resulting in lessened endochondral ossification, repressed proliferation of chondrocytes in growth plate cartilage, diminished cellular hypertrophy and decreased cartilage matrix production; leading to a mixture of clinical manifestations and complications. The gene has been diagrammed to band 4p16.3. In the heterozygous state, achondroplasia is non-lethal, with most patients having a fairly normal life span and normal intelligent quotient. The children affected with condition are short at birth and grow slowly throughout childhood; the average final height for women is 123 cm and 130 cm for men.(9)

Characteristic clinical features are classically present at birth; with affected patients having short limbs, a long narrow trunk and a large head with midfacial hypoplasia and a prominent forehead. The rhizomelic limb shortening and the fingers shows trident configuration. Most joints are hyperextensible, but extension is restricted at the elbow is evident. Several odonto-stomatological manifestations of achondroplasia were skeletal and dental class III malocclusion, a narrow maxilla, macroglossia and an open bite between the posterior teeth .(9)

The case of achondroplasia can be diagnosed prenatally as following :

- Prenatal ultrasound –During third trimester , shortening of limbs and hydrocephalus can be detected
- Aminocentesis – In this procedure ,the amniotic fluid which surrounds the fetus is studied to examine the FGFR3 mutation
- Chorionic villus sampling – The placental tissue is been tested ,to detect mutation in FGFR3 gene (10)

The case of achondroplasia can be diagnosed postnatally as following :

PHYSICAL EXAMINATION

- Unusually short thighs and arms
- Prominent forehead with a small upturned nose
- Small chest
- Broad, flat feet ,short toes, short fingers
- Extra space between middle and ring fingers called a trident hand
- Weak muscle tone
- Bowed legs

RADIOGRAPHIC EXAMINATION

- Lateral skull which reveals midface hypoplasia ,enlarged calvaria, shortening of base of skull ,foramen magnum is diminished in size
- AP view shows narrowing of interpedicular distance
- CT to calculate lung volumes

- MRI reveals fusion of C1 , cervico-medullary compression (11)
- Sleep study to observe airway obstruction
- Pulmonary function test to check the vital capacity

In our case we diagnosed through conventional radiographs like PA SKULL,OPG and HANDWRIST though super diagnostic modalities of radiograph like CT,MRI can be done for accurate understanding of the present case

In hypochondroplasia ,the condition is characterized by short stature, stocky built, mild joint laxity ,other skeletal features are similar as seen in achondroplasia but tend to be medical complications like spinal stenosis ,tibial bowing ,obstructive apnoea occurs less frequently in hypochondroplasia .Epilepsy and intellectual disability but pertaining to this case, she is not epileptic and her IQ is normal ,thus hypochondroplasia is excluded(4)

In chondrodysplasia ,people with chondrodysplasia classically have normal intelligence and a normal life expectancy but some serious life threatening complications including stenosis of airways which restricts breathing ,abnormalities of heart ,above mentioned findings are not evident in this case(5)

1.GROWTH HORMONE

This is aided using somatotrophin (recombinant growth hormone). To start treatment for ACH with human GH, the patient must satisfy all of the following selection criteria :

(i) Standing height body measurement for children of age 3years

(ii) Bone age is one of the criteria where boys should be 17years and girls should be 15 years old

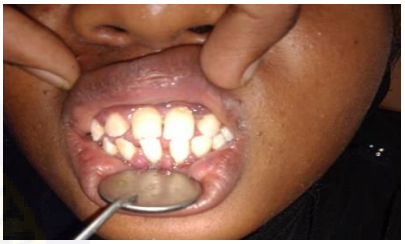
(iii) The current date height of the body is 3 SD below standard height compared to children of the same sex and age;

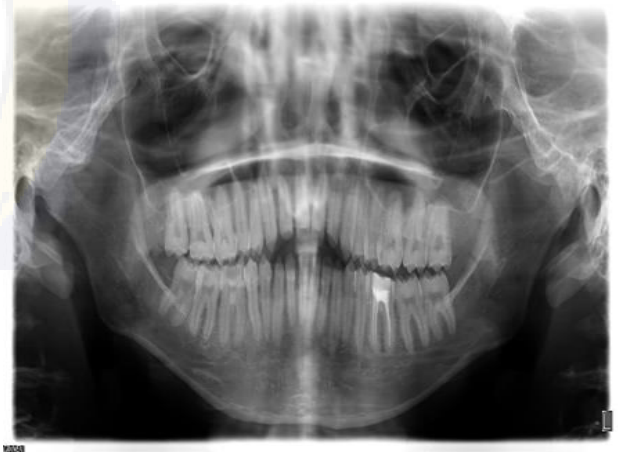
(iv) The patient should display the physical features of achondroplasia

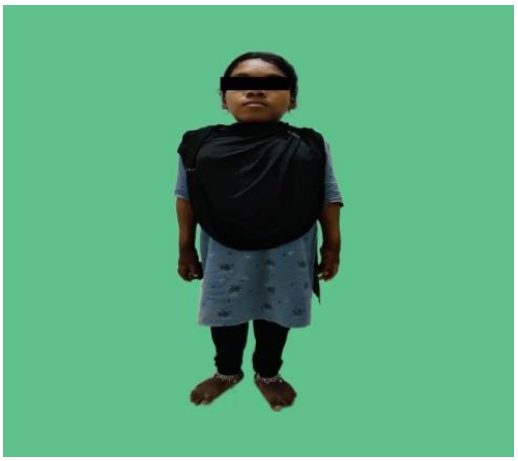
(v) An MRI , CT examination should not reveal complications such as foramen magnum stenosis, lumbar spinal canal stenosis, hydrocephalus, and spinal compression sufficient to permit surgery. Neurological manifestations due to compression are not seen to proceed the treatment. (12)

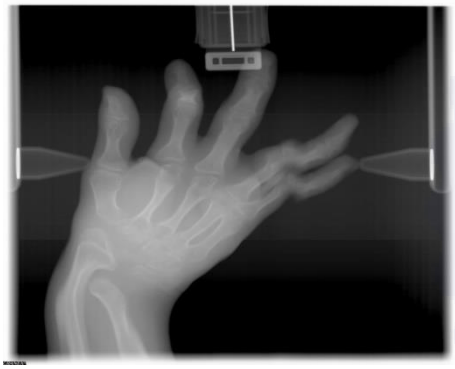
2. LIMB LENGTHENING:

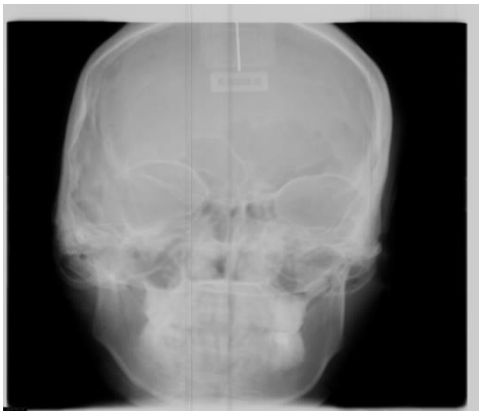
This procedure is done to increase length of limbs and to increase the stature. External fixators are helping in limb lengthening, but it will be long term process the recommended age for doing this procedure is patients older than 12 years(13)











CONCLUSION

Growth is a complex process in which nutrition, hormones, genetic factors, and environmental factors play important roles. Recent findings in molecular and cellular biology as well as clinical genetics have highlighted a wide group of other mechanistical systems that regulate skeletal growth and an accompanying vast array of genetic disorders that can cause defects in normal growth. Dentist, especially those working in child and maternal health, may be required to intervene in cases of short stature. Adequate knowledge on the underlying conditions, their treatment, and the need for counseling can greatly improve patient outcomes and prevent the development of short stature and assist in improving the quality of life of individuals who have already attained full maturation. These efforts require an interprofessional healthcare team that includes the family clinician/dentist, specialists like endocrinologist ,pediatrician etc,.

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ACKNOWLEDGEMENT: NIL

TABLE 1: SALIENT FEATURES IN EACH GIVEN CONDITION AND THEIR ASSOCIATED DISEASE

SALIENT FEATURES	DISEASES
Frontal bossing	Osteogenesis imperfecta, Rickets, Acromegaly, Cleidocranial dysplasia, Gardner syndrome, Cogenital syphilis, Crouzon syndrome, Ectodermal dysplasia, Hallerman steriff syndrome β thalesemia
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Midfacial hypoplasia	Pierre robin syndrome, Fetal alcohol syndrome Achondroplasia, Crouzon syndrome
Trident hand	Achondroplasia, Anauxetic dysplasia ,Hypochondroplasia

CHART 1 : FAMILY HISTORY OF THE PATIENT

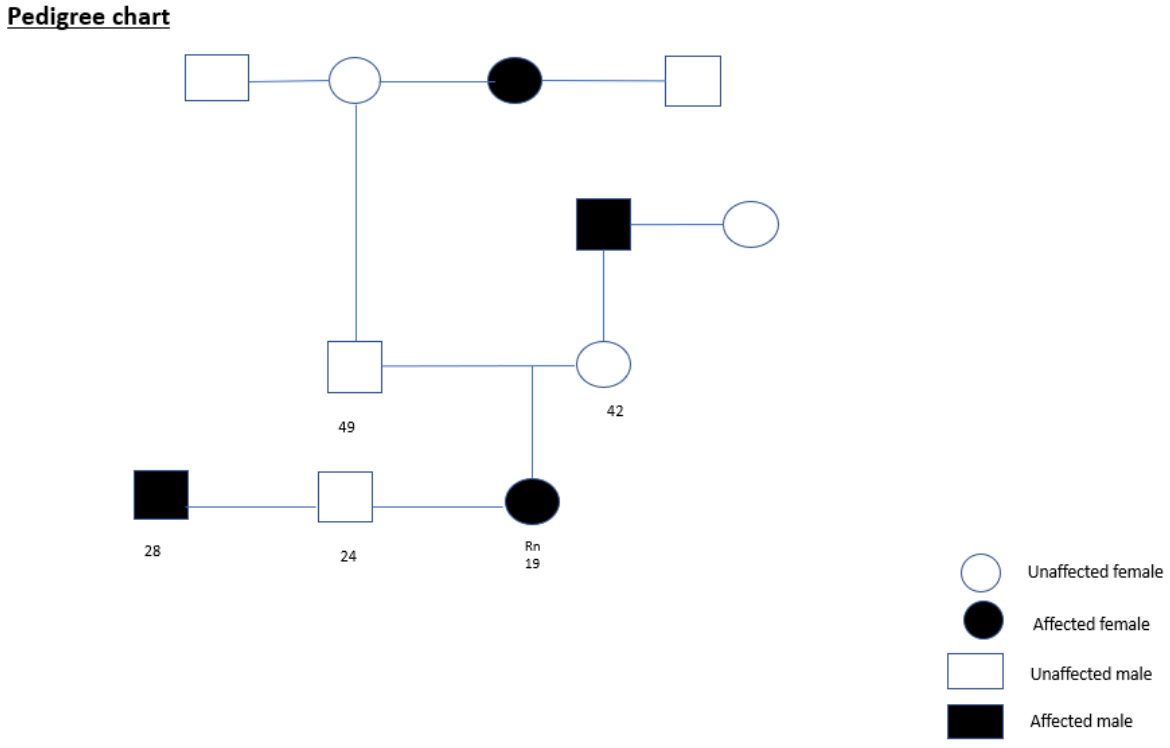
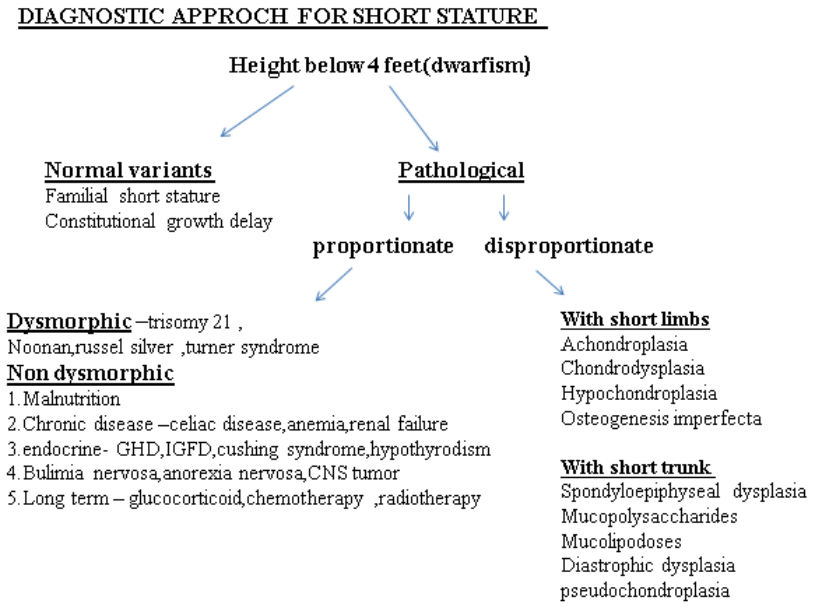


CHART 2: DIAGNOSTIC APPROACH FOR SHORT STATURE



IMAGES

FIGURE 1 : CLINICAL PICTURE OF THE PATIENT SHOWING SHORT STATURE , RHIZOMELIC SHORTENING OF ARMS AND LEGS

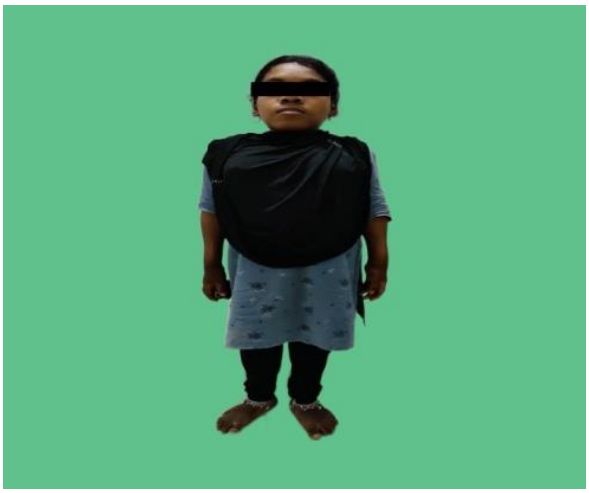


FIGURE 2: FINGERS SHOWING TRIDENT HAND COFIGURATION

FIGURE 3: INTRAORAL PICTURE

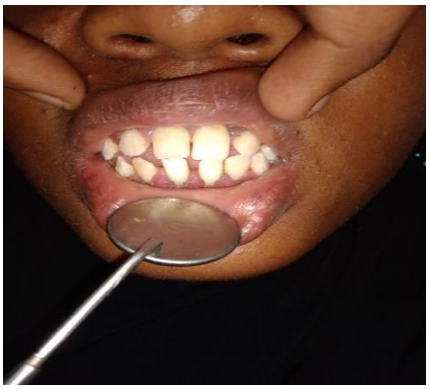


FIGURE 4 OPG OF THE PATIENT

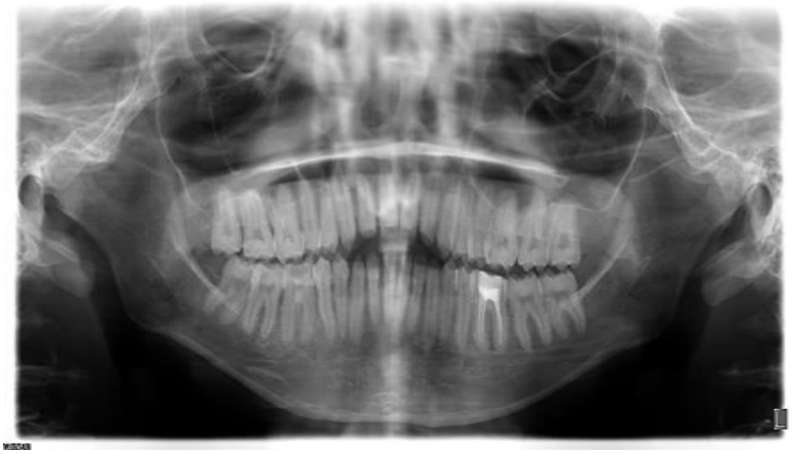


FIGURE 5: PA VIEW SHOWING ENLARGED CLAVARIA

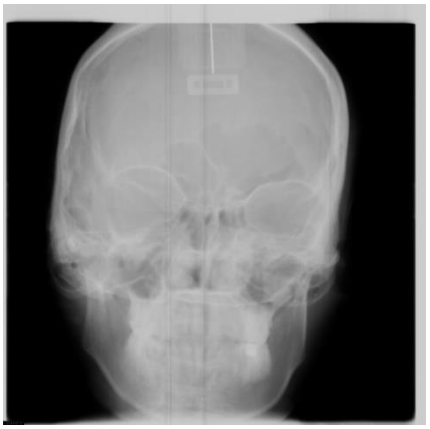


FIGURE 6: TRIDENT CONFIGURATION OF HAND