Case Report

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KENNY-CAFFÉY SYNDROME TYPE 2 AND GORLIN – CHAUDHARY-MOSS SYNDROME: A RARE CASE WITH DENTAL MANIFESTATIONS

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ABSTRACT:

Kenny–Caffey syndrome is a rare hereditary skeletal syndrome.KCS Type 2 is an autosomal dominant form caused by mutations in FAM111A gene, characterized by growth retardation, short stature, uniformly small slender long with medullary stenosis, thickened cortex of the long bones, hypocalcemia and normal intelligence. Gorlin- Chaudhary-Moss syndrome is an extremely rare inherited syndrome, which is inherited as an autosomal recessive trait, and is characterized by brachycephaly, microphthalmia, hypodontia, and hypertricosis. Affected individuals also have growth retardation and also mild mental retardation. We present the case of a 26 year old male with Kenny–Caffey syndrome Type 2 and Gorlin-Chaudhary-Moss syndrome with emphasis on the craniofacial and dental manifestations and the treatment options.

Key Words: Kenny – Caffey syndrome type 2, Gorlin-Chaudhary-Moss syndrome, hypodontia, microdontia,hypocalcemia, hypoparathyroidism,growth retardation.

INTRODUCTION:

Kenny-Caffey syndrome type 2 is an autosomal dominant rare hereditary skeletal syndrome caused by mutations in FAM111A, with normal intelligence. It affects the skeleton, head and the eyes. It is characterized by delayed closure of anterior fontanel and abnormally large head circumference with a prominent forehead. Occular abnormalities include microphthalmia and papilledema. Hypocalcemia is seen within two months after birth. Hypocalcemia is caused by hypoparathyroidism due to improper functioning or due to absence of parathyroid glands.

Gorlin-Chaudhary-Moss syndrome is an extremely rare autosomal recessive syndrome characterized by premature closure of the sutures between the skull leading to brachycephaly, microphthalmia, hypodontia, and hypertrichosis. Individuals also have growth retardation and mild mental retardation. In addition, maxillary hypoplasia and abnormally narrow and high arched palate is also noticed. Dental abnormalities includes hypodontia, microdontia, peg shaped, abnormally shaped and positioned teeth leading to difficulty in mastication.

CASE DETAIL:

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A 26-year-old, male bank employee by profession reported to the Department of Oral Medicine and Radiology, Pushpagiri College of Dental Sciences, Thiruvalla, Kerala South India, with the chief complaint of missing upper and lower teeth. His main concern was his esthetics and difficulty in mastication. Medical history details revealed Kenny Caffey syndrome and Gorlin-Chaudhary-Moss syndrome.

He had low serum calcium levels of 7 mg/dl, phosphorous level of 7.4 mg/d, raised alkaline phosphatase levels of 207 U/L and low PTH level of 7pg/ml. Growth hormone levels were as low as 1.8ng/ml. Patient was on Inj. Humatrope 0.4mg daily for 5 months and stopped as the clinical response to the GH therapy was not good. Patient is on Tab Shelcal -500mg, Tab Alfacip -0.25 mg. Hemogram, liver, renal function, urine analysis and thyroid hormones were within the normal limits. Skeletal survey of skull and hands revealed, pituitary fossa appeared large, anterior clenoids were indistinct, the cranial fossa appears deep and the convolutional markings were prominent. The bones of the extremities appeared normal except for minimal increase in bone density. Chromosomal analysis was done (GTG-banding) indicating a male karyotype. Suspects an interstitial deletion in 22q. Karyotype-46, XY.

RCT of 46 and extraction of 43 were done previously and it was uneventful. No history of consanguineous marriage. He was born as the first child at full term by normal vaginal delivery. His birth weight was 2.3 kg and his brother is anormal adult.

General oral examination revealed short stature around 126 cm tall and 29 kgs of weight. His intelligence was normal. Fig 1 shows the stature of the patient and Fig 2 shows the digits of the right and left hand.

Extraoral examination revealed frontal bossing,high frontal hair line, minimal scalp hair, thin lips, long philtrum, micrognathia, deep set eyes,microphthalmia, ear lobules low set and slender bones. Fig 3 shows extraoral photograph of the patient, Fig 4. shows the microstomia and Fig 5 shows lateral profile of the patient.

There was maxillary hypoplasia, abnormally narrow, high-arched palate, microstomia and hypodontia. Teeth present are 11, 13, 52 root stump, 16, 17, 21, 22, 23, 64, 65, 73, 74, 36, 37, 41, 46 and 47. Peg lateral and microdont in relation 22 and talons cusp in relation to 21. Teeth are abnormally positioned leading to difficulty in mastication. Remaining teeth were missing. Fig 6 Intraoral photograph showing the maxillary arch, Fig 7 shows the mandibular arch and Fig 8 showing Malocclusion.

Panoramic radiograph revealed multiple missing teeth with no impacted permanent tooth bud, incomplete root formation of the centrals and thin marrow cavities with medullary stenosis. Mandible appears to be small with small coronoid and condyles. (Fig 9 Panoramic radiograph, Fig 10 A-P view).

A final diagnosis of Kenny-Caffey syndrome and Gorlin-Chaudhary-Moss syndrome with craniofacial and dental manifestations was made with these findings.

A comprehensive treatment plan was advised with full mouth rehabilitation that includes 1. Extraction of 11, 21, 14, 64, 65, 73, 74, 41, 46, 47.

2. Intentional RCT of 13, 23, 16, 17.

3. Maxillary and mandibular overlay dentures.

Patient was referred to the departments of prosthodontia, oral surgery and endodontia, where the following treatment is being carried out for the restoration of esthetics, form and function of the masticatory apparatus to near normalcy.

DISCUSSION:

Kenny–Caffey syndrome was first reported by Kenny and Linarelli in 1966.[1] Caffey described its radiological features in 1967.[2] Lee described the classical facial features in 1983.[3] Recurrent episode of hypocalcemia and low levels of phosphates in the blood. Affected individuals exhibit short stature height ranging from 48-59 inches. Intelligence is usually normal.[4,5,6] Males and females are equally affected. Individuals have thickened cortexes of the long bones and abnormally thin marrow cavities. The anterior fontanel is abnormally large, closes late so the affected infants have an abnormally large head with a prominent forehead. They have small eyes, papilledema, and hypermetropia.[7,8]

Diagnosis is confirmed by x ray studies of the skeleton showing distinctive thickening of the outer layers of the long bones with unusually thin marrow cavities. Blood tests detect episodes of hypocalcemia.

Related disorders includes Kenny–Caffey syndrome type 1, also called hypoparathyroidism–retardation dystrophic syndrome (HRD) caused by disruption of the tubulin–specific chaperone E (TBCE).[9-14] Characterized by hypoparathyroidism at birth, growth retardation, intellectual disability and facial abnormalitieslike deep set eyes, thin lips, micrognathia and depressed nasal bridge. Individuals also have abnormally thin marrow cavities and hypocalcemia.

Osteopetrosis is a rare genetic skeletal disorder characterized by the abnormal thickening of the bones, delayed closure of anterior fontanel and anemia.

Pycnodysostosis is a rare disorder characterized by short stature and increased density of bone. Individuals have short arms and legs with broad hands and feet with delayed closure of the fontanels. Other features include macrocephaly with small face, small jaw, brittle finger and toe nails, blue sclera and dental abnormalities.

Vitamin D and calcium are effective in treating hypocalcemia and iron supplements for anemia. Dental abnormalities has to be treated for

maintaining proper functioning and esthetics. Ophthalmology check is necessary for treating the eye abnormalities. Prevention achieved through preimplantation genetic diagnosis and carrier detection.

Gorlin-Chaudhary-Moss syndrome is a congenital condition manifested in an autosomal recessive manner also called as craniofacial dysostosis. Exact etiology remains unknown.[15] The first case was reported in 1960, characterized by growth retardation, short stature, and mild mental retardation.

Due to craniosynostosis, brachycephaly is present. Midface hypoplasia, maxillary hypoplasia and abnormally high arched palate are the other features. Dental abnormalities like hypodontia, microdontia[16] and teeth are abnormally positioned causing malocclusion and difficulty in mastication. Hair line may be low on the scalp and the affected infants have hypertricosis.

Skeletal abnormalities includes hypoplasia of the bones at the end of the fingers and toes and also of the metacarpals leading to short hands, fingers and toes. Females may show underdeveloped genitalia.[17]

No specific diagnostic techniques other than ruling out conditions that show similar signs and symptoms. Complications include astigmatism, farsightedness, ductusarteriosus.[18]

Related disorders includes Weill-Marchesani syndrome a rare inherited disorder of the connective tissue characterized by brachycephaly, brachydactyly, cataracts, and glaucoma. Saethre-Chotzen syndrome is a rare inherited disorder characterized by craniosynostosis, brachycephaly, midface hypoplasia, malformed ears, high arched palate, maxillary hypoplasia, extra, missing or peg shaped teeth. Individuals also exhibit brachydactyly and syndactyly.[18]

Treatment of Gorlin-Chaudhry-Moss syndrome require coordinated efforts of a team of pediatricians, surgeons, cardiologists, ophthalmologists, dental specialists, speech specialists, ENT specialist to systematically and comprehensively plan child's treatment. Surgery may be performed to correct craniofacial abnormalities. Partial or complete dentures, extractions and dental restoration can be done.

To prevent this syndrome, genetic testing of expecting parents and prenatal diagnosis using molecular testing of the fetus may help in understanding the risk during pregnancy. If there is a family history of the condition then genetic counselling will help assess risk before planning for a child. Quality of the life can be improved through close monitoring of the condition and by addressing the signs and symptoms appropriately.

CONCLUSION:

Most of the PLS patients were identified and diagnosed by dental professionals. Hence thorough knowledge about this syndrome may help the dentist to make a diagnosis at an early age. If left untreated,

PLS may lead to edentulism at an early age which makes early diagnosis & intervention necessary. Proper oral hygiene maintenance and flap surgeries with bone grafts and Platelet Rich Fibrin as early as possible have given better results.

The role of periodontists is more in treating patients with Papillon Lefevre Syndrome, awareness about this syndrome should be made through education and conducting awareness programs.

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FIGURES:

Fig1: Stature of the patient

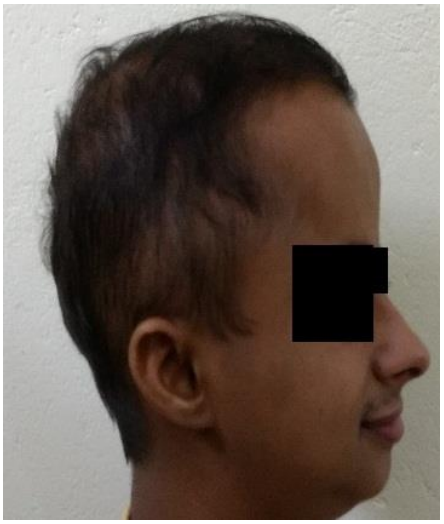

Fig 3: Extraoral lateral photograph

Fig 2: Extraoral Frontal photograph

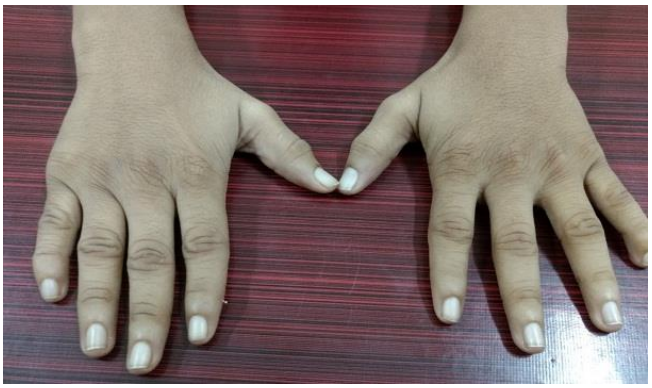

Fig 4: Photograph of the digits of the patient

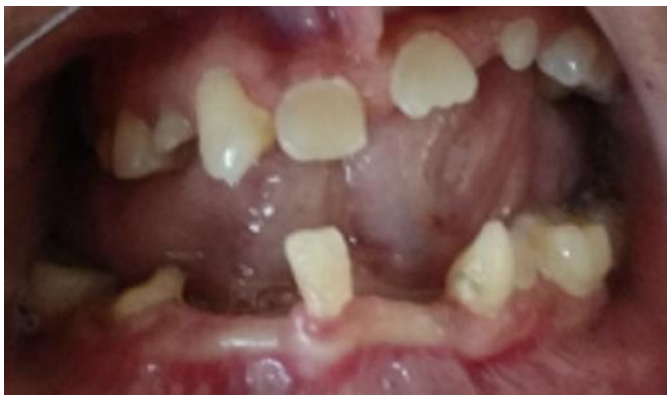

Fig 5: Intraoral photograph showing microstomia

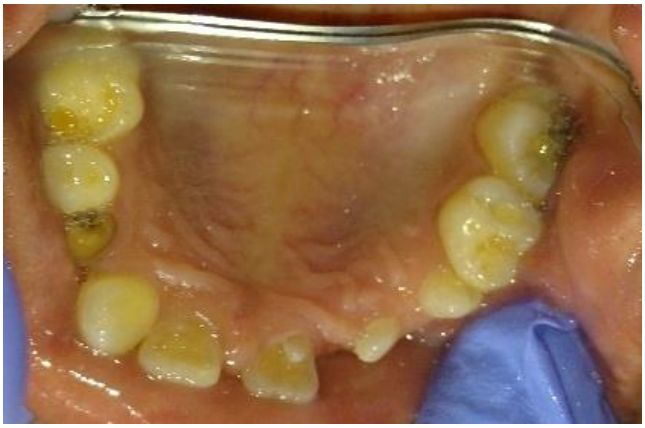

Fig 6: Intraoral photograph of the maxillary arch

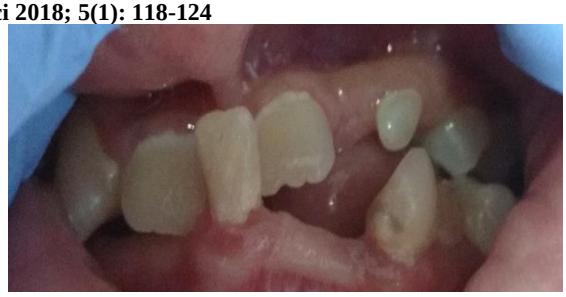

Fig 8: Intraoral photograph showing Malocclusion

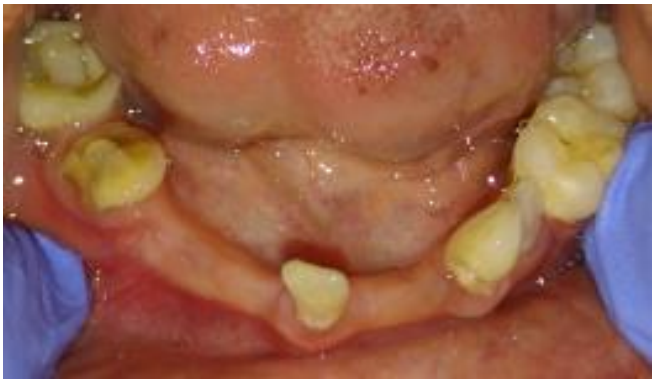

Fig 7: Intraoral photograph of the mandibular arch

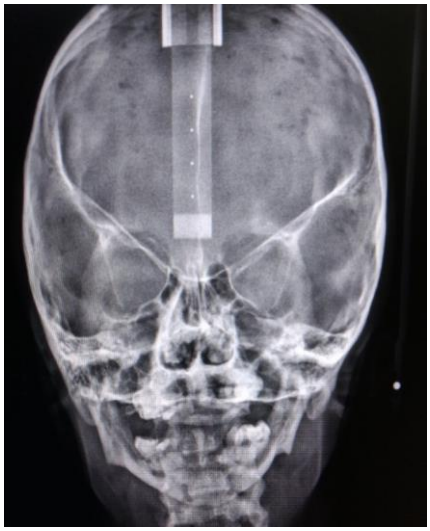

Fig 10: A-P View

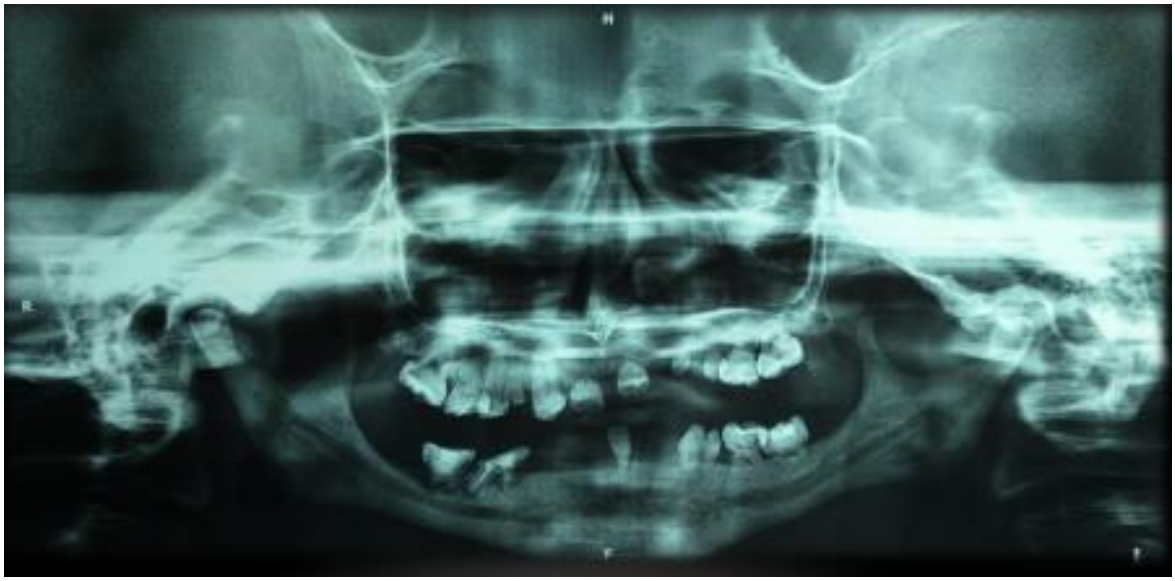

Fig 9: Panoramic radiograph