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CASE REPORT

GORDON SYNDROME: A CASE REPORT

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ABSTRACT: BACKGROUND: Gordon Syndrome is an extremely rare disorder and it is part of a group of genetic disorders known as Arthrogryposis multiplex congenita. There are congenital contractures in at least two or more areas of body. Gordon Syndrome is characterized by congenital Camptodactyly, Clubfoot and Cleft Palate. **CASE CHARACTERISTICS:** One month female baby presented with congenital distal contractures in hands (Camptodactyly), talipes equinovarus deformity in both lower limbs (Club feet), hyperextension of left leg at knee joint, central cleft palate, oral thrush. **INTERVENTION:** Treatment of oral thrush, feeding advice and physiotherapy. **OUTCOME:** Relieved of oral thrush, weight gain started and attached to Plastic Surgery, Orthopedic and Physiotherapy Specialties. **MESSAGE:** Sporadic cases of this rare disorder do occur and not many cases have been reported from India.

KEYWORDS: Arthrogryposis, Cleft Palate, Camptodactyly, Talipes.

INTRODUCTION: Gordon Syndrome has been indexed as a rare disease in the database of NORD (National Organization for rare diseases),[1] and also in Genetic and rare disease Information center.[2] The term is synonymously used with Distal Arthrogryposis Type IIA, Camptodactyly-Club foot-Cleft Palate, Arthrogryposis Multiplex Congenita Distal type IIA. Recently it has been classified as Arthrogryposis Distal type 3; (DA3). It is more of a clinical finding than a disease. Arthrogryposis derives its name from Greek, literally meaning "Curving of joints". Arthron meaning "joint" gruposis meaning "hooking". It is supposed to be genetically transmitted as an Autosomal dominant trait or as single mutation. It affects males and females equally. Characteristic physical features are obvious at birth manifesting as stiffness or contractures with impaired mobility of hands and feet along with deformity of bones, permanent fixation of fingers in flexed position (Camptodactyly), abnormal positioning of fingers and toes with medial rotation and inward position of feet (Talipes) and other malformations of extremities, with cleft palate. Affected child may also show other deformities and abnormalities of bones and joints.

CASE REPORT: One month female child was brought with history of bent feet, cleft palate since birth and difficulty in feeding for 2 days. The child belonged to healthy Nepali parents residing in Himachal Pradesh for 2 years. There was no consanguinity in marriage, no family history of deformity or defects on either paternal or maternal side. However mother was a third para who had an elder normal daughter of 5 years and 5 months and had lost her 1st born male child who was still born. All the children were born by normal vaginal delivery at home. The family belonged to a poor socioeconomic class.

On examination the baby was afebrile, conscious with normal vital signs. There was no icterus, clubbing, cyanosis, pallor, oedema, or lymphadenopathy. There was oral thrush, her head circumference was 36 cm, weight 2460 gm, length 49 cm. There was mild hypertelorism and central cleft palate (Photo annexure1) mild talipes equinovarus deformity in left foot and marked talipes

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equinovarus deformity in right foot (CTEV). Right knee was normal while left knee showed marked genu valgus deformity with anterio medial rotation of fibula (Photo Annexure 2). Both hands were fixed in a state of hyperflexion and there was subluxation of both thumbs at metacarpophalangeal joints. Other Systems were normal.

Investigations revealed a normal haemogram with CPK –MB 26.0 IU/L and CPK.

Total 252.0 IU/L levels (normal for age).

X-ray lower limbs showed bony equinovarus deformity of right foot and genu valgum deformity at right Knee. X-ray chest, USG abdomen, CT head and echocardiogram were normal.
She was diagnosed as having Gordon Syndrome (Arthrogryposis multiplex congenital Type II A).

DISCUSSION: The triad of camptodactyly, cleft palate and club foot was earliest reported by H Gordon in 1969.[3] He first described the syndrome in a family showing autosomal dominant transmission through three generations. Thereafter no reports appeared till 1979 when Fahed Halal reported a second family with a pattern consistent with autosomal dominant inheritance with reduced penetrance in females through 5 generations.[4] Not many cases have been reported in Indian literature. Gordon Syndrome as seen in present case is a very rare congenital syndrome, manifesting with distal contractures and deformities of multiple joints and cleft palate. Fetal akinesia has been attributed to be the main precipitating factor.[5] Prenatal diagnosis by ultrasonography is possible by7 to 8 weeks when fetal movements can be made out by USG[6] and there lies the importance of antenatal checkups. In the present case mother did not go for any antenatal checkups and had not received any medicines or nutritional supplementations. There can be marked variability in expression of disease both intra and Interfamilialy. Therefore careful assessment is very important for prognostic counselling and rehabilitative management as well as genetic counseling.[7]

Other defects like drooping of eye lids (ptosis), micrognathia or retrognathia or defects in vertebrae might also be associated.

Gordon syndrome with congenital myopathy has been recently reported from South Africa.[8] It is supposed to be caused by heterozygous mutations in the PIEZO2 gene on18p11 chromosome.[9] Recently it has been classified as Arthrogryposis distal type 3 (DA3).[10]

The causative factors leading to fetal akinesia are supposed to be many and can be classified as, extrinsic – where there is insufficient space in uterus for normal fetal movements, like abnormal shape of uterus, fetal crowding or insufficient amniotic fluid and intrinsic - muscular, neurological, or connective tissue disorder in the fetus. Joint development is always normal but since motion is essential for further normal development of joints and their contiguous structures, lack of fetal movements causes contractures. Treatment is directed towards specific symptoms present in each case requiring a coordinated effort from different specialties.

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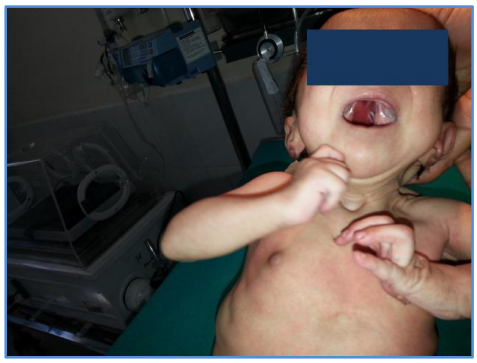

Fig. 1: Cleft Palate

Fig. 2: Arthrogryposis

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