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”Cloves Syndrome”-Report of A Rare Case with Lipoma of the Conus As An Additional Feature

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Abstract: *“CLOVES” stands for Congenital Lipomatous Overgrowth, Vascular malformations, Epidermal naevi, Spinal or Skeletal anomalies. CLOVES Syndrome is extremely rare ,non hereditary, progressive overgrowth disorder, with about 100 cases reported worldwide. The features of this syndrome include truncal lipomatous lesions, cutaneous naevi, vascular anomalies (typically truncal) and spinal anomalies like Scoliosis or skeletal anomalies like limb overgrowth, widened first interdigital space, overgrowth of digits etc. Some are also associated with seizures or central nervous system malformations like hemimegalencephaly. This recent delineation distinguishes it from Proteus syndrome, a disorder that comprises localized, progressive, postnatal overgrowth with bony distortion, dysregulated adipose tissue, cerebriform connective tissue and linear epidermal nevus, hemimegalencephaly, and other manifestations. The author describes the case of a 9 months old child who presented with features of CLOVES syndrome with an additional finding of a lipoma of the conus which has not been reported so far.*

Keywords: *Cloves syndrome,Truncal lipomatosis,vascular malformation,tethered cord*

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I. Introduction

“CLOVES” stands for **C**ongenital **L**ipomatous **O**vergrowth**,V**ascular malformations**,E**pidermal naevi**,S**pinal or **S**keletal anomalies.CLOVES Syndrome has been recently described,is an extremely rare ,non hereditary progressive overgrowth disorder,with about 100 cases reported worldwide.The features of this syndrome include truncal lipomatous lesions,cutaneous naevi,vascular anomalies(typically truncal) and spinal anomalies like scoliosis or skeletal anomalies like limb overgrowth,widened first interdigital space,overgrowth of digits etc.Some are also associated with seizures or central nervous system malformationslike hemimegalencephaly.We herein report a case of a child with features of CLOVES syndrome,with a Lipoma of the conus as an additional feature,which has not been reported so far.

II. Materials and Methods

A 9 months old female child presented with progressively enlarging swelling in the trunk.The swelling was not present at birth but was first noticed by the parents when the child was three months old.She was the first child, born at 40 weeks, to non consanguinous parents.Antenatal history was unremarkable.On examination she was found to have a large soft globular swelling in the trunk(fig.1).There was a Port wine stain over both buttocks and sacral region(fig.2). X-rays did not reveal any skeletal anomaly.MRIof the spine revealed a lipoma of the conus with tethered cord(fig.3).HRCT of the thorax revealed a lipohaemangiomatous growth in the mediastinum(fig.4).MRI of the brain was normal.A diagnosis of CLOVES syndrome was arrived at on the basis of truncal lipomatous overgrowth,epidermal naevi,mediastinal vascular malformation and spinal anomalies of tehered cord and conus lipoma.

III. Results

Patient was operated upon by a team of Neurosurgeon and a Plastic surgeon,the conus lipoma was excised and de-tethering of cord was done,followed by near total excision of the truncal lipoma.Postoperative period was uneventful and the child was discharged after 2 weeks. At One and half years follow-up,the child has normal bladder bowel function with normal power in both lower limbs,but has developed regrowth of the truncal lipomatous mass(fig.5) Two years after surgery,patient presented with paraparesis and bladder bowel retention of sudden onset.MRI of the Dorsal spine showed a the lipohaemangiomatous growth in the mediastinum had enlarged in size and extended into the spinal canal through the left D6 vertebra,compressing the cord.Also the MRI showed evidence of a thrombosed vessel on the surface of the intraspinal component of

the lipohaemangiomatous mass.The mediastinal mass and the intraspinal extension were excised in two separate surgeries.At,6 months follow-up after the second surgery,patient has regained bladder bowel function and is ambulatory with support.

IV. Conclusion

Cloves syndrome is a very rare, progressive disorder affecting multiple organs with no specific guidelines for its treatment.Long term prognosis is still unclear and much more data and experience are required in this respect.A multidisciplinary approach is required for the treatment of this condition.

V. Discussion

The pathogenesis of CLOVES has been identified. Kurek et al[4] identified mutations in PIK3CA in six patients with CLOVES syndrome, and mutant allele frequencies ranged from 3% to 30% in affected tissue from multiple embryonic lineages. They conclude that CLOVES is caused by postzygotic activating mutations in PIK3CA[5].Further studies are needed to support these mutations. We could not study this mutation in our patient as genetic analysis facilities are not available in our hospital or any other centre in the vicinity.

The most prominent features reported by Alomari et al[1],in their cohort of 18 patients with CLOVES syndrome were truncal lipomatous masses of variable size, complex vascular malformations (including lymphatic, arteriovenous, and phlebectasia), scoliosis, and skeletal and other anomalies. In addition to the vascular anomalies, there is a wide spectrum of imaging findings in CLOVES syndrome reflecting the asymmetric body overgrowth and musculoskeletal and other internal organ anomalies. Musculoskeletal findings include extremity bony and soft tissue overgrowth, leg length discrepancy, chondromalacia patellae, dislocated knees, scoliosis, wide triangular feet with widened first interdigital space or large hands, and macrodactyly, typically involving the third toe or third finger, talipes, and neural tube defect.The lipomatous overgrowth characteristic of CLOVES syndrome shows a tendency to recur post surgery.[1]

Central nervous system (CNS) involvement is also described in this syndrome. The anomalies include partial agenesis of corpus callosum,a four layered cortex, polymicrogyria, non-contiguous abnormalities of the gray and white matter, and ventriculomegaly[3]. Neural tube defects and tethered cord were also described by Saap *et al*.[6] and Alomari[2].Our patient had a Lipoma of the Conus, in addition to low lying tethered cord,which has not been reported till date. Management of CLOVES syndrome requires a multidisciplinary team of Neurosurgeon,Plastic surgeon,dermatologist and Paediatrician.Increased risk of Pulmonary embolism is described in patients with CLOVES syndrome[2].Longterm prognosis in these patients is unknown as data and experience in treating this condition is limited due to its rarity.

Conflict of Interest:The author declares there is no conflict of interest in presentation of this paper.

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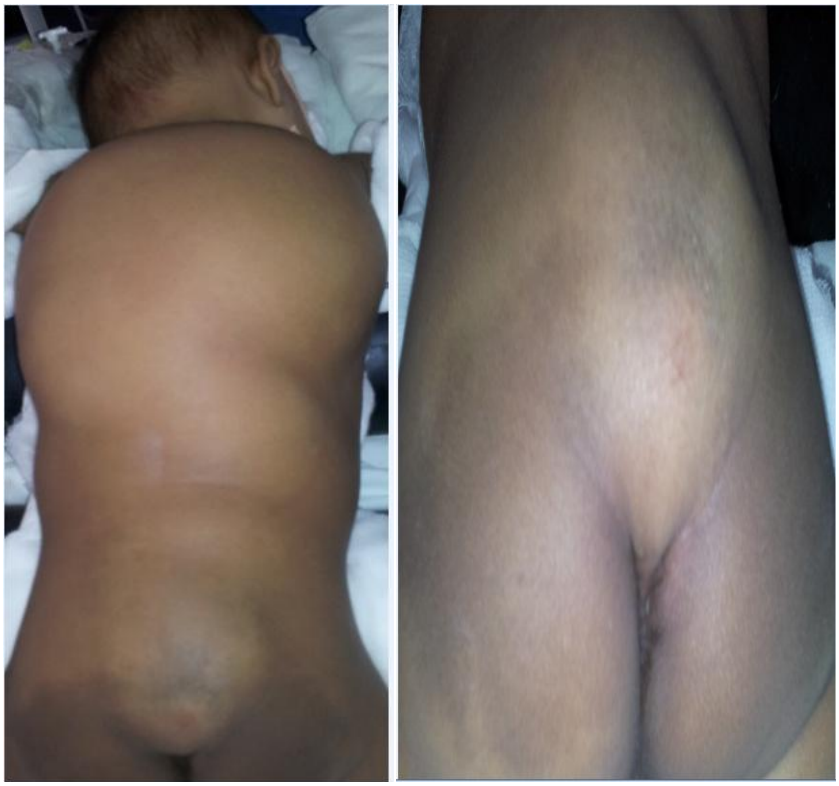

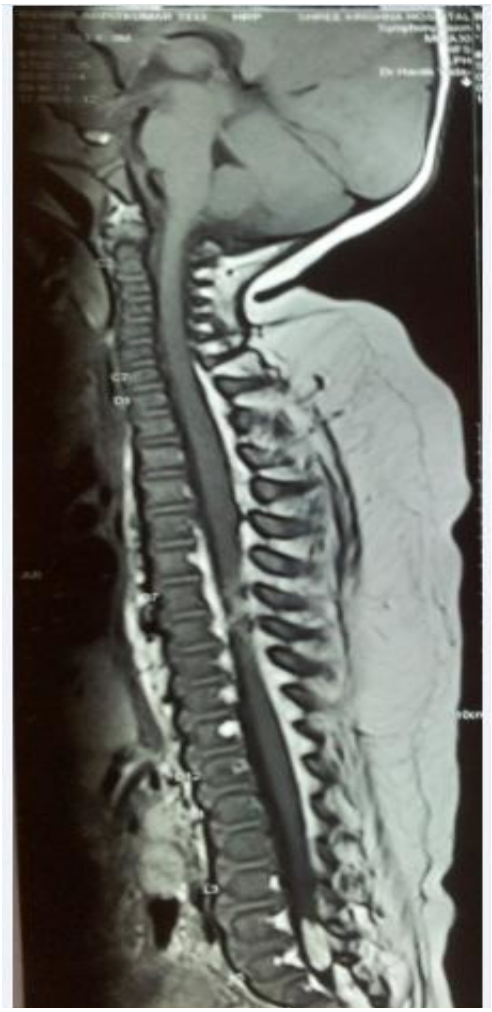

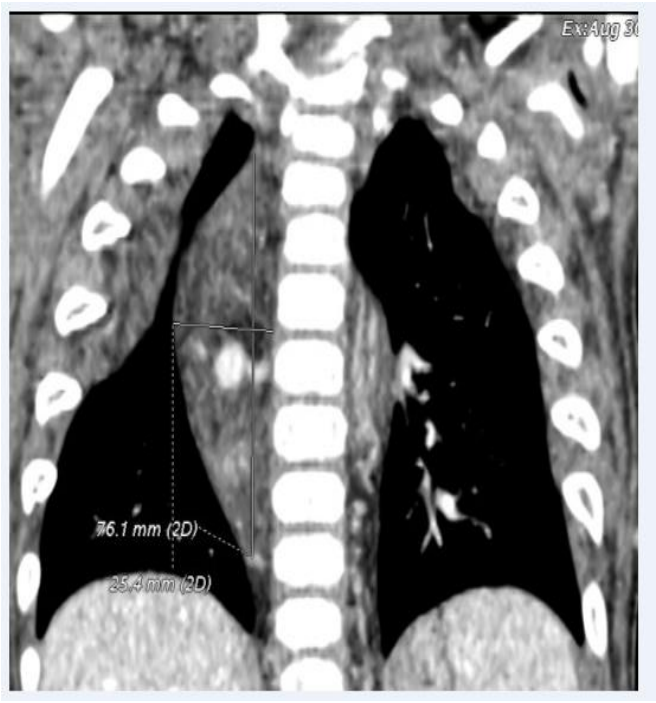

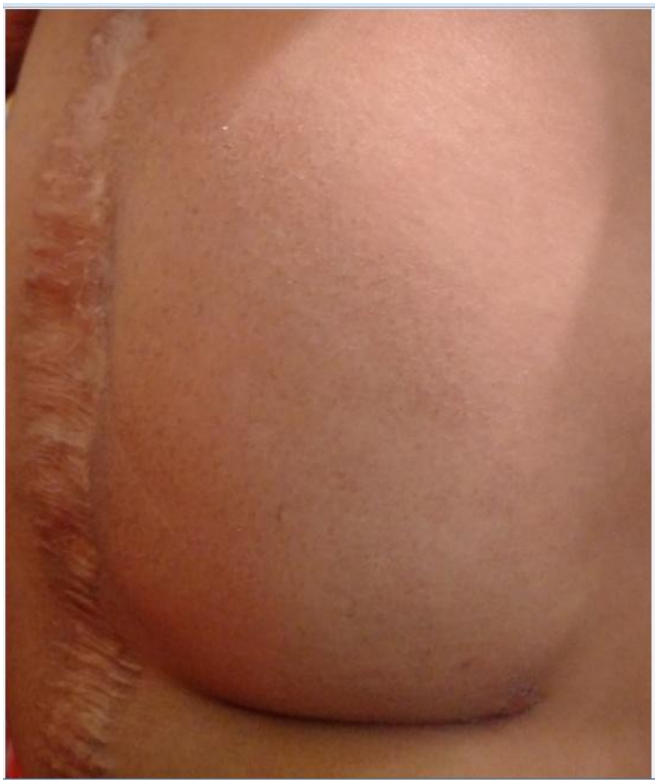

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