Original Research Paper

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General Medicine

PROTEUS SYNDROME WITH SPLENIC CYSTS

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ABSTRACT Complex hamartomatous Proteus Syndrome was characterized by multiple diverse somatic manifestations. We present a case with skeletal, soft tissues, skin and vascular lesions but with unusual rare finding of splenomegaly with multiple cysts, sparing the Central Nervous System. On a careful review of Indian literature on the subject, there have been no prior documentation of Proteus Syndrome with splenic cysts.

KEYWORDS : PROTEUS SYNDROME-MULTIPLE SPLENIC CYSTS:

Proteus Syndrome is a complex hamartomatous disorder first described by COHEN & HAYDEN in 1979. It was so named by Rudolf Wiedemann et al (German Pediatrician) in 1983 after the Greek God Proteus, who could change his form at will. This syndrome has multiple diverse, somatic manifestations that evolve over time and involve the skeletal system, soft tissues, skin and vascular system2, resulting in partial gigantism of the hands, feet, asymmetry of the limbs, plantar hyperplasias, macrodactyly, bony exostosis, soft tissue tumours (hemangioma, lymphangioma, lipoma), varicosities, verrucous epidermal nevi and long bone over growth1 .

CASE REPORT:

18 year old female of non consanguinous from Krishna District of Andhra Pradesh with abdominal lump, large size of right foot and swelling on back of right chest since birth, which were progressive in nature. On examination she was moderately built, anaemic with stable vital data, height 57 inches, arm span length 61 inches, drooping of right shoulder with kyphosis is seen. An obliquely placed soft tissue swelling of 11 inches x 9 inches present on the right back of chest extending to axilla and right lumbar region with multiple dilated veins over the swelling (Fig. 1 & 2). Abdominal examination revealed an irregular swelling on the whole left side of abdomen extending to right lumbar and iliac regions, with irregular margins and nodular irregular surface moving with respirations; firm to hard in consistency, dull on percussion, nontender, no local increase of temperature, no bruit heard over. Multiple dilated veins, papular rash and multiple bluish soft subcutaneous nodules seen over the swelling (Fig.2). Umbilicus is everted, and transversely split. A large sized right great, second and third toes were seen in triradiate position (Fig. 4). Clinically no abnormality in Cardiac, Respiratory and Central Nervous Systems. Biochemistry showed Hb 8 grams, normal liver function tests, normal blood sugar and urea. Blood examination revealed microcytic hypochromic anemia with atypical lymphocytes.

Ultrasound of abdomen showed high positioned liver, splenomegaly (23 cm x 11.3 cm) with multiple cystic lesions (Fig.8). Left kidney compressed by enlarged spleen posterolaterally. Right kidney and both ovaries were normal. X-ray of right foot showed enlarged first, second and third metatarsals and tarsal bones. X- ray chest (PA & Rt Lat) showed (Fig.7) soft tissue density retrosternally and posterior mediastinal mass. ECG, Fundus, MRI of brain were normal. Chromosomal studies were normal.

DISCUSSION:

Postulated mechanisms of its occurrence –

Proteus Syndrome results from lethal somatic gene mutation, which leads to mosaic state that allows its survival. Receptors of tissue growth factors may be altered causing multiple, diverse, somatic manifestations that evolve over time and involve the skeletal system, soft tissues, skin and vascular system. These signs include partial gigantism of the hands and or feet, asymmetry of limbs, plantar hyperplasia, macrodactyly, bone exostosis, soft tissue tumours (haemangioma, lymphangioma, lipoma), varicosities, verrucous epidermoid nevi and long bone over growth. Palmar and plantar gyriform masses are considered as pathogonomic sings, histologically these are connective tissue lipomas.

Spleen is a reticuloendothelial organ, which develops from the dorsal buds of dorsal mesogastrium at about 5 weeks of gestation and later fuses in dorsoventral direction and occupies the adult location i.e., left upper quadrant. The remaining dorsal mesogastrium divide into lenorenal and gastrosplenic ligaments.

The ectodermal reminent cells in the dorsal region may have lineage of mutated gene for proteus growth factors, which can give rise to cysts embedded in spleen in the course of its development. They are more common in females than males and usually found in childhood or adolescence [3].

Non –neoplastic cysts can be divided into true (primary) cysts, which posses a cellular lining and false (secondary) cysts which have no cellular lining. True cysts are either parasitic (echinococcal) or non parasitic (epithelial). True cysts may be lined by a specific secreting membrane, these are epithelial (dermoid, epidermoid), endothelial (lymphangioma, hemangioma, polycystic, serous). 80% of cases of congenital splenic cysts are unilocular and solitary. It has been suggested that they are derived from inclusions of the mesothelial lining of the splenic surface on the splenic parenchyma during development. The growth of splenic cysts to the parenchyma of the lining cells or accelerated secretion from these cells. Further growth can be attributed to bleeding from the cystic wall, as well as to an osmatic imbalance of the cystic fluid like that which occurs with other cystic lesions.

CONCLUSION:

Proteus Syndrome should be considered in the differential diagnosis of hamartomatous and neurocutaneous syndromes. So far reported cases[2,3] revealed cystic lesions in Brain and Lung, in our case there was huge Splenomegaly with multiple cysts.

Figure- 1 Increased pad of fat in the upper 1/3 of plantar aspect of Right foot and triradiate toes

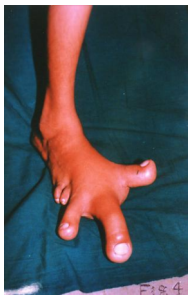
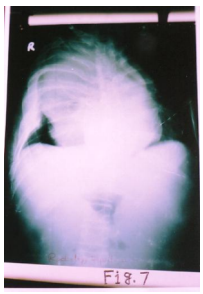


Figure- 2

X-rays of chest- PA view shows mass in mediastinum and soft tissue density retrosternally and post mediastinal mass.



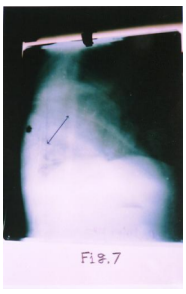
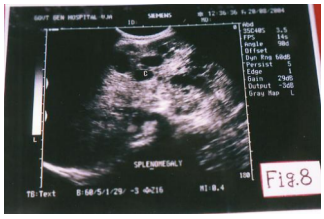


Figure- 3

Ultra sound of abdomen showing Splenomagaly
(23 Cms x 11.3 Cms) with multiple cystic lesions – 'C'



Figures 1 & 2

Shows 11" x 9" swelling on Right back of chest with multiple dilated veins.

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