PICTORIAL

SKIN LESION IN MUCCOPOLYSACCHARIDOSIS (MPS) TYPE II: HUNTERS DISEASE

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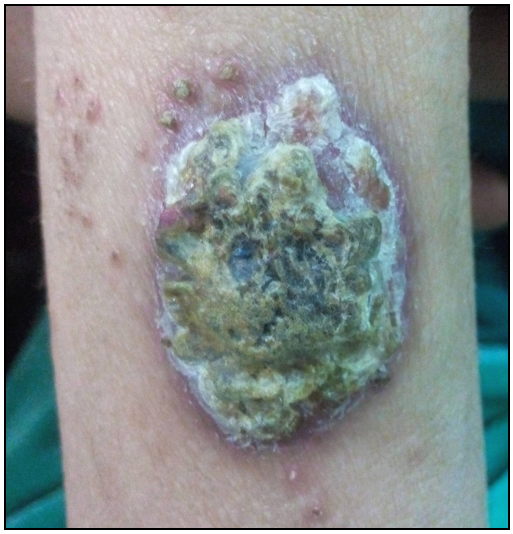

A 12 year old boy presented with short stature, coarse facial features, stiff joints, clear corneas and mental retardation. X-ray imaging showed Cardiomegaly, characteristic bony features called Dysostosis multiplex comprising of tapering, bullet shaped metacarpals and phalyngeal bones, and oar shaped ribs and ovoid vertebral bodies. There were multiple skin lesions present on trunk and upper limbs. These were ivory coloured, with characteristic pebbly appearance. Spot urinary GAG was increased, confirming the diagnosis of Muccopolysaccharidosis (MPS) type II also called Hunter Disease.

MPS are inherited disorder, and unlike rest of types type II, Hunters disease, is X-linked disorder. Treatment modalities available are enzyme replacement therapy and Hematopoietic Stem Cell Transplant.[1]

Skin lesions in MPS are very rare; still it can be the earliest sign of Hunters disease in mild cases. MPS II patients can present with distinctive skin lesion (pebbling). Biopsy can be taken from this lesion which will show deposits of Glycosaminoglycans.[2]

REFERENCES

1 Muenzer J, Overview of the mucopolysaccharidosis, Rheumatology (2011) 50 (suppl 5): v4-v12.

2 Martin R, Beck M, Eng C, Giugliani R, Harmatz P, Mufioz V, *et al.* Recognition and diagnosis of mucopolysaccharidosis II (Hunter syndrome) Pediatrics. 2008;121:377–86.

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