Infantile galactosialidosis associated with extensive Mongolian blue spots: An uncommon presentation

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Sri Lanka Journal of Child Health, 2023; **52**(2): 236-238
DOI: https://doi.org/10.4038/sljch.v52i2.10349
(Key words: Galactosialidosis, inborn errors of metabolism, Mongolian spots, Genetic analysis, Benign manifestation)

Introduction

Mongolian spots are common benign skin marks present at birth, which are hereditary or developmental in nature[1]. However, extensive Mongolian spots involving large areas of the back, trunk, and extremities merit special consideration as they can be an early marker of certain inborn errors of metabolism (IEM) like GM1 gangliosidosis, Hurler syndrome, mucolipidosis, mannosidosis, Niemann-Pick disease and galactosialidosis[2,3]. A few case reports are available, which refer to instances of IEM associated with extensive Mongolian spots. Here, we describe a case of infantile galactosialidosis with extensive Mongolian spots.

Case report

A 6-month-old boy, born to 2nd degree consanguineous healthy parents was admitted following concerns of developmental delay. He was found to have coarse facial features including broad nasal bridge, a long philtrum, and frontal bossing (Figure 1), but no corneal opacities or macular cherry red spots. He had hepatosplenomegaly, hypotonia with brisk tendon reflexes and pedal oedema.

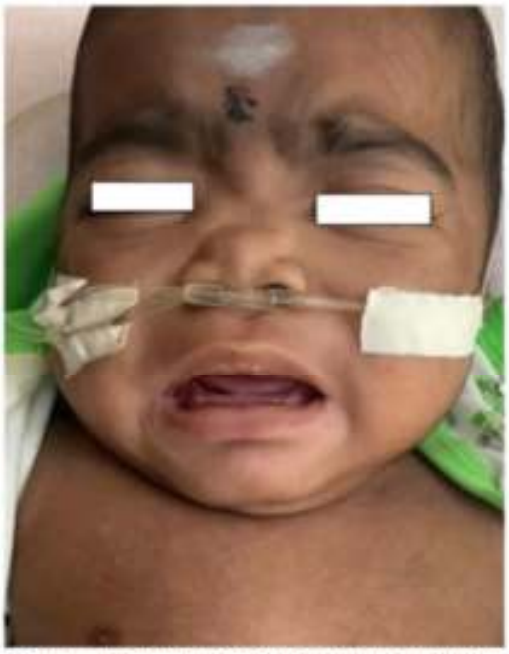

Figure 1: ***Coarse facial features including broad nasal bridge and a long philtrum***
**Permission given by parents to publish photograph*

Neonatal records revealed hydrops fetalis after birth. In addition, large hyper-pigmented, well demarcated patches resembling Mongolian blue spots were scattered all over the body sparing face and upper limbs (Figure 2).

Figure 2: ***Extensive Mongolian pigmentation***

There was hypertrophic cardiomyopathy on 2D echocardiography. Ultrasound scan of the abdomen showed hepatosplenomegaly with normal kidneys and normal portal pressure. Radiography revealed anterior beaking of the lower part of the vertebrae and oar shaped ribs (Figure 3).

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(Received on 09 October 2022: Accepted after revision on 18 November 2022)

The authors declare that there are no conflicts of interest
Personal funding was used for the project.
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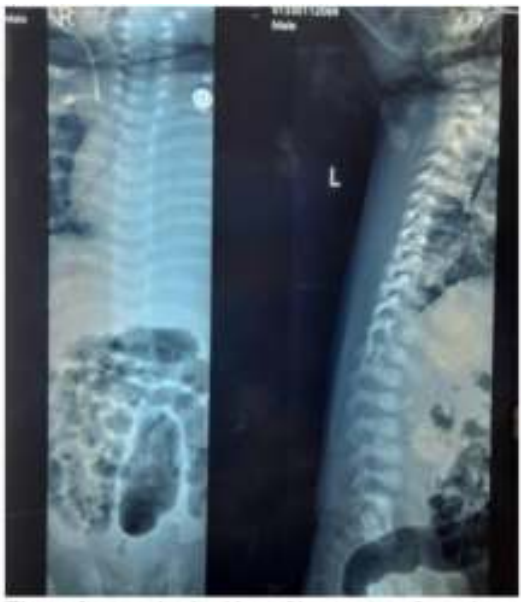

Figure 3: ***Anterior beaking of lower part of vertebrae and oar shaped ribs***

His liver synthetic functions, uric acid levels, lipid profile, amino acid profile, urine organic acid levels and urine glycose amino glycan levels were normal. Genetic analysis done through next genome sequencing (NGS) - based copy number variation (CNV) analysis identified the cystatin A (CTSA) variant c.1045>A p.(Cys349Ser).This variant has previously been described as possible galactosialidosis by Kostadinov S, *et al*[4].

Hence, with the clinical findings and the presence of CTSA variant, a diagnosis of infantile galactosialidosis was made.

Discussion

Galactosialidosis is a rare, autosomal recessive, lysosomal storage disorder. It results from defects in glycoprotein degradation due to mutation in a single gene, encoded by the protective protein cathepsin A (CTSA), located on chromosome 20q13.12[5]. It is classified into three types based on age of onset and clinical phenotype. Infantile phenotype is characterized by developmental delay, hypotonia, visceromegaly, inguinal hernias, skeletal changes and ocular abnormalities which develop between birth and 3 months of age. Death occurs at an average age of 8 months, usually from cardiac or renal failure[5].

In addition to these clinical findings, our patient had extensive Mongolian blue spots which were scattered over the trunk and lower limbs, often anterior in location, as typically seen in lysosomal storage disorders[6]. There are 54 reported cases of extensive Mongolian blue spots with various IEM, in which 25 cases are associated with Hurler syndrome, 17 with GM1 gangliosidosis, 9 with Hunter syndrome, 2 with alpha–mannosidosis and 1 with Niemann-Pick disease[7].

To conclude, Mongolian spots should not always be considered benign; it can be a decisive factor for the early identification of lysosomal storage disorder especially in the context of developmental delay. Even though there is lack of curative treatment, early identification is important to provide family planning and early palliative care decision.

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