From mongolian spots to mucopolysaccharidosis

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Background

Mucopolysaccharidosis VI or Maroteaux-Lamy syndrome is a lysosomal storage disease caused by a deficient activity of the arylsulfatase B (ASB) enzyme. This results to a progressive accumulation of glycosaminoglycans (GAGs) into lysosomes and extracellular matrix, with subsequent cell and tissue injuries and a serie of multi-systems/organs failure, leading to severe clinical manifestations[1]. MPS VI is very rare, with incidence estimates ranging from 0.36 to 1.30 per 100,000 live births[2].

Case description

A 2-month-old baby boy with multiple episodes of respiratory infections and a history of cardiomyopathy presented for consultation with multiple mongolian spots on his arms, legs and buttocks since birth. We also notice facial dysmorphism.
The demonstration of an abnormally high quantity of urinary dermatan sulfate as well as a genetic test highlighting the presence in the homozygous state of the variant c.971G>T, p.Gly324Val in the *ARSB* gene made it possible to confirm the diagnosis of MPS VI. Our patient was able to benefit from a replacement enzyme treatment.

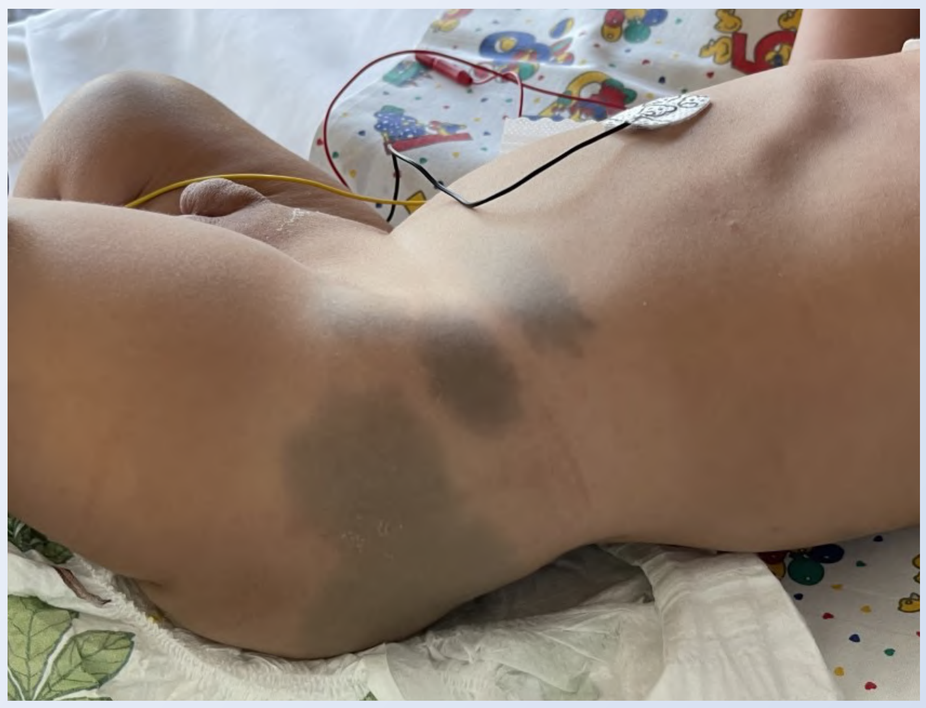

Fig.2 : multiple ectopic mongolian spots

Fig.1 : Facial dysmorphism

Discussion

MPS type VI is characterized by facial dysmorphism and a short stature accompanied by other progressively worsening symptoms between 6 and 24 months: joint limitations, very severe dysostosis multiplex, hepatomegaly, heart valve damage, cardiomyopathy, spinal cord compression, meningeal thickening, communicating hydrocephalus, deafness, corneal opacities. Intellectual development is usually normal, but auditory and ophthalmological impairment can cause learning disabilities.
The average life expectancy is 30 years. Death occurs from cardiac and respiratory complications.
The differential diagnosis are other forms of mucopolysaccharidosis (types I and II) or oligosaccharidosis[3]. The treatment is based on enzyme replacement therapy (galsulfase) to improve endurance ans cardiorespiratory profile[4].

Conclusion

MPS type VI is a rare disease but should be considered in the presence of a child presenting with ectopic mongolian spots with or without facial dysmorphism.

References

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