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Sengers syndrome-A rare cause of hypertrophic obstructive cardiomyopathy

Manjusha Hira and Emanuel Quist Therson
Watford General Hospital, UK

Introduction: Sengers syndrome is a rare disorder that causes congenital cataract, hypertrophic cardiomyopathy (HOCM), skeletal myopathy and lactic acidosis. HOCM is usually fatal in infancy. It is an autosomal recessive mitochondrial depletion disorder resulting from the mutation of acylglycerolkinase (AGK) gene. This nuclear gene is responsible for the maintenance of mitochondrial DNA (mtDNA).

Methods: We report a 4 month old boy who had severe lactic acidosis at birth and progressive congenital cataract. Cataract and failure to thrive were missed on his primary examination until he presented with complete loss of vision at 3 months and subsequent heart failure secondary to severe HOCM at 4 months of age.

Results: His genetic testing revealed homozygous novel putative splicing mutation of AGK gene, c.1047-2A>G. This substitution has not been previously reported. There are about 40 case reports of Sengers syndrome to our knowledge. The severe form due to homozygous mutation has early onset cataract, lactic acidosis and progressive HOCM leading to death in infancy. The milder form with heterozygous mutation has a better prognosis and develops cardiomyopathy at later stages, with survival into their fourth decade. The mildest form reported had only cataracts.

Conclusion: Uncertainty about the red eye reflex in non-Caucasian infants is common in primary examinations. Unexplained and persistent lactic acidosis at birth should not be discounted and should be followed up after discharge. Sengers syndrome should be considered as a differential diagnosis in babies who present with congenital cataracts especially if associated with lactic acidosis and later HOCM. Our patient is currently awaiting a cardiac transplant.

Biography

Manjusha Hira is a Senior Registrar in Pediatric at Watford General Hospital UK .She has published more than 6 papers in reputed journals.

msha201409@gmail.com

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