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Legius syndrome in a 13 month old boy: A case report

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Background: Legius syndrome is autosomal dominant and caused by mutations in the SPRED1 gene. Clinical manifestations include multiple cafe-au-lait spots, axillary/inguinal freckling and a degree of macrocephaly, without the non-pigmentary signs of neurofibromatosis type 1 (NF1). Learning disabilities, developmental delay and ADHD are known. It is a rare disorder (fewer than 200 individuals with a confirmed diagnosis), and difficult to differentiate from NF1 in early childhood. This is important in terms of prognosis and monitoring. We describe a case in a 13 month old boy.

Case: A 13 month old boy was referred by the GP with a one day history of a dilated right pupil on a background of a viral URTI for which parents were giving nasal decongestant spray. His neonatal, developmental and past medical history was unremarkable. His head circumference was on 99.6th centile. The eye signs resolved spontaneously, thought to be due to the nasal spray. On assessment he was noted to have multiple cafe-au-lait spots >5 mm in diameter. Cafe-au-lait spots were also noted on his mother and brother. The rest of his clinical examination was unremarkable. Baseline bloods including metabolic screen as well as MRI brain were normal. His initial neurofibromatosis type 1 analysis was negative. He went on to have further genetic screening which revealed 'heterozygous for SPRED1 c.229A>T', confirming a diagnosis of Legius syndrome.

Conclusion: This is a rare disorder and difficult to differentiate from NF1, highlighting the importance of identifying more affected individuals to better delineate the clinical picture and course.

Biography

Dipali Shah is a Consultant Pediatrician at West Middlesex University Hospital, UK. She is a Pediatric Assessment Unit Lead. Her interest is Under-graduate and Post-graduate education, training and teaching. She is working as a Consultant since November 2013. She has also worked as a Quality Improvement Fellow. She has done four poster presentations.

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