Case Report

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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 also 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. The prevalence of Legius syndrome is unknown. Many individuals with this disorder are likely misdiagnosed because the signs and symptoms of Legius syndrome are similar to those of neurofibromatosis type 1.

Other typical NF1 features such as Lisch nodules of the iris, neurofibromas and central nervous system tumors are systematically absent .Two studies revealed that approximately 2% of individuals fulfilling diagnostic criteria for NF1 have SPRED1 mutations.

Case: A 13 month old boy was referred by the GP to our pediatric assessment unit; with a one day history of a dilated right pupil on a background of a viral upper respiratory tract infection for which parents were giving Xylometazyline Hydrochloride nasal spray for two days. He was born at tem via c-section. There was nothing significant in the past. His development was appropriate for his age. There was no family history of epilepsy.

His head circumference was on 75-91%centile. On assessment he was noted to have multiple café-au-lait spots >5mm in diameter. They were 17 in number. There were 9x5 cm Café-au-lait spots noted on his chest. The café-au-lait spots were also noted on mother's arm and brother's body. The rest of the examination was unremarkable. There was no hepatospenomegaly.

At this point neurofibromatosis type1, tuberous sclerosis, Horner's syndrome, posterior fossa tumor, dilated pupil due to the nasal spray were considered as differential diagnosis.

His baseline bloods including metabolic screen and MRI brain were normal. The eye signs resolved spontaneously next day, thought to be due to the nasal spray. 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.

He was arranged for the outpatient review. He was also referred to the geneticist for the family screening and counseling. But parents did not attend. Subsequently three more appointments were sent, still parents did not bring him and we lost the follow up.

Other Conditions with Café-au-lait spots are fanconi's anaemia, neurofibromatosis type 1 like syndrome, tuberous sclerosis, silver Russell syndrome, multiple neoplasia type 1, McCune–Albright syndrome etc.

Treatment is symptomatic. For attention deficit and hyperactivity drug treatment is recommended. Physiotherapy, occupational therapy as well as speech therapy is considered for the developmental delay. Education support is required for the learning disability children. The genetic counselling is must.

Conclusion

This is a rare disorder and difficult to differentiate from NF1, highlighting the importance of identifying more affected individuals to better define the clinical presentation and course [1-3].

References

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