

Case Report

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Tuberous Sclerosis Presenting as Delay in Development of Speech

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Abstract

A 3-year-old male child presented as delay in development of speech, a rare single presenting feature with tuberous sclerosis. Clinical history and examination were consistent with tuberous sclerosis including major features like Ash-leaf spots and Adenoma sebaceum. The clinical manifestations and evaluation of tuberous sclerosis are discussed.

Keywords: Tuberous sclerosis; Speech; Developmental delay

Introduction

Tuberous sclerosis was first described in eighteen hundred eighty by Bourneville (a French physician). The most frequently involved organs are brain, kidneys, lungs, heart, skin and skeleton. A combination of symptoms may include seizures, developmental delay, behavioural problems, skin abnormalities, and lung and kidney disease.

About 50% of people with Tuberous Sclerosis have learning difficulties ranging from mild to significant forms [1]. Studies have reported that 25% to 61% of affected individuals meet the diagnostic criteria for autism, with an even higher proportion showing features of a broader pervasive developmental disorder [2]. A recent study reported self-injurious behaviour in 10% of people with tuberous sclerosis [3]. Other conditions, such as ADHD, aggression, behavioural outbursts and OCD have also been reported. Selective involvement of delay in speech as a presenting complaint has not been reported in literature. We report an interesting case of a 3 year old male child, who came to our out-patient department with sole complaint of delay in speech and on examination skin manifestations of tuberous sclerosis were detected accidently, leading to further work up and subsequent diagnosis of the same.

Case

A male child aged 3 years presented to us with complaints of delay in development of speech. The child could only speak monosyllables while his other milestones were normal for his age. On detailed history his mother told that he runs, climbs upstairs with alternate stepping, can easily open the lids of containers and even open the knob of the door. The birth history was uneventful and there was no history suggestive of any prenatal asphyxia; Post natal period, newborn and infancy were normal too. There was no history of any CNS insult and the child achieved rest of the milestones normally. There was no history of any irritability and any other behaviour abnormality and no cognitive impairment or any evidence of pervasive developmental disorder. There was no family history of seizures or any delay in developmental milestones in elder siblings. No history of any prolonged illness or prolong intake of any medications was present.

On examination all the anthropometric indices were normal for age. The child had normal I.Q (as per Sanguin Form Board test). On general examination mild pallor was noticed, multiple papular eruptions (Adenoma sebaceam) were seen on face (Figure 1) and 3-4 hypopigmented lesions over the back (Ash leaf spots) (Figure 2) were present. The parents were not sure about the time of appearance of the lesions over the body.

No other neuro-cutaneous marker was seen especially over the

lumbar area and there was no evidence of any periungual fibromas. No dental pits or gingival fibromas were seen. Central nervous system examination was normal without any behavioural abnormality. On investigation, his MRI Brain showed multiple bilateral cortical tubers, involving mainly the right parietal lobe; with numerous small tubers seen in rest of the areas. The ventricular system was normal without any evidence of dilation. On fundoscopy retinal hamartoma was seen. His echocardiogram and ultrasound abdomen was normal and there was no evidence of rhabdomyoma and angiomyolipoma in heart or kidneys respectively. Genetic test was not done, as the child was fulfilling the clinical criteria. Further, screening of other siblings revealed no evidence of Tuberous Sclerosis.

Discussion

Although learning difficulties and features of autism are present in cases of Tuberous Sclerosis but there is no reported case presenting only as delay in development of speech. Further, development of adenoma sebaceum was quite early in the patient. The diagnosis was suspected on the basis of hypopigmented lesion on the trunk and was



Figure 1: Patient with Adenoma Sebaceum on face.

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Received March 07, 2013; Accepted April 12, 2013; Published April 15, 2013

Citation: Abqari S, Anjum A, Khan DI, Nawab T, Sardha M, et al. (2013) Tuberous Sclerosis Presenting as Delay in Development of Speech. J Clin Case Rep 3: 269. doi:10.4172/2165-7920.1000269

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Figure 2: Patient having hypopigmented (Ash-leaf spots) patches on his trunk.



confirmed into "definite" category by fundus examination and MRI Head (Figure 3).

Although, focal or generalized seizures with or without developmental delay are often the first sign of the disease, but careful examination can detect hypo-pigmented lesions even earlier, providing an early diagnostic clue [4]. Tuberous sclerosis still has varied presentation in different forms, as reported previously [5].

Delay in speech as an isolated sign in TSC is quite unusual; we don't have clear explanation for this. Similarly early development of adenoma sebaceum is equally intriguing. Longer follow up is needed to see if the progression of the disease is rapid in this particular case, so far there was no episode of seizures and rest of developmental milestones were according to age.

Conclusion

A high index of suspicion should be kept as Tuberous Sclerosis in patients of development delay, especially those not presenting with its classical clinical features (e.g. seizures etc.). Further, all efforts should be made to detect any neuro-cutaneous markers in such cases.

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