عنوان مقاله:

Always consider Prader-Willi Syndrome: case report

محل انتشار:

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خلاصه مقاله:

BACKGROUND: Prader-Willi syndrome (PWS) is genetic disorder caused by loss of function of chromosome 15 q11-q13. It can cause wide range of symptoms including neonatal hypotonia, low birth-weight, hypogonadism, cognitive impairment and poor sexual developments. Before age 3, the most important symptom is hypotonia and then other symptoms become apparent.CASE PRESENTATION: Here we report 1-year-old Iranian boy (46, XY) who was the first child of consanguineous parents and was referred to us for evaluation of hypotonia, poor sucking and abnormal genitalia. Examination revealed hypotonia, bilateral undescended testis(UDT), small testes, micropenis, small hands, almond-shaped eyes, blond hair. He had been visited for many times and had taken many tests but there was no significant diagnosis. We noticed to his history and after the careful examination he was suspected for PWS and was referred to genetic test which indicated heterozygous deletion and confirmed PWS.CONCLUSION: No specific biomarker is yet available for PWS and confirmation would be by genetic test. It is diagnosed in approximately one in 25,000 births. Starting growth hormone therapy (GH treatment) under the age of years may improve language skills and cognition and motor development. So an early diagnosis of PWS can be so helpful therefore, although the prevalence of PWS is not high and most of the symptoms appear after years clearly, considering PWS as differential diagnosis would be good idea especially when the clinical symptoms are UDT, hypotonia and some facial features like strabismus and almond-shaped eyes.

کلمات کلیدی: