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A Rare Syndromic Case of Hypocalcaemia

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Introduction

Congenital hypoparathyroidism-retardation-dysmorphism (HRD) has been described by Sanjad in 1988. It is a rare autosomal recessive disorder mainly reported from the Middle East. It is characterized by congenital hypoparathyroidism, both growth and mental retardation and with distinctive dsymorphic features. It can be confused with other syndromes presenting with hypocalcaemia and dysmorphic features.

Objective

To raise awareness of Sanjad Sakati Syndrome in describing the following case who presented with hypocalcaemia and severe psychomotor retardation.

Case Report

A girl presented at the age of 4 months with fever and was found to be hypocalcaemia. Her growth parameters were markedly below 3rd centile. Phosphate level was high and her parathyroid hormone level was low. She had distinctive dysmporphic features; microcephaly, deeply seated eyes, thin lips, beaked nose, micrognathia, small hands and feet. Furthermore she was hypotonic and had developmental delay. The girl was diagnosed as a case of Sanjad-Sakati syndrome. The girl was managed with Calcium supplements and alphacalcidol. However, the management of such cases is challenging in balancing the control of calcium and phosphate levels and the adverse effects of treatment as well as managing the growth delay.

Conclusions

The triad of hypoparathyroidism, severe failure to thrive and distinctive dysmporphic features should lead to early recognition and proper management of such a challenging condition as Sanjad Sakati. It can sometimes be confused with Kenny-Caffey Syndrome, Di George Syndrome and familiar hypoparathyroidism Genetic studies are available to confirm the diagnosis and antenatal counselling.