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Pallister-Hall syndrome: Case report

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An 8 year old boy was brought with history of developmental delay and seizures since 6 months of age. He was the first child of a third degree consanguineous couple. He had been born full term and had cried immediately at birth. He had been detected to have penoscrotal hypospadias at birth for which he had undergone multiple corrective surgeries. On examination, he had dysmorphic facies with bulbous nose tip, small stubby hands with central polydactyly and short stature. The MRI of the brain showed a hypothalamic hamartoma. Pallister-Hall syndrome is characterized by hypothalamic hamartoma, central polydactyly and variably other abnormalities including imperforate anus, bifid epiglottis and panhypopituitarism. It is inherited as an autosomal dominant, however many cases arise as a result of spontaneous mutations. It is associated with frame-shift mutations of the *GLI3* gene. Patients with Pallister-Hall syndrome are reported to have easier seizure control and lesser incidence of precocious puberty as compared to patients with isolated hypothalamic hamartomas.

Biography

H. Bhasin has her expertise in evaluation and passion in improving the health and wellbeing of children. She has been working in Kalawati Saran Children's Hospital, India

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