Congenital hypopituitarism associated with complex cranio-vertebral junction anomalies: a case report

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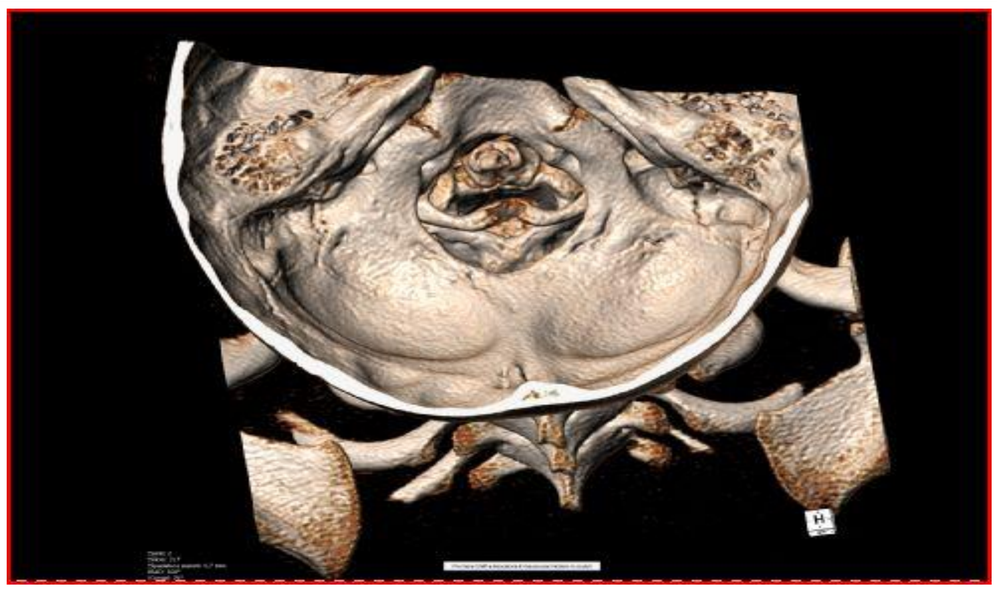
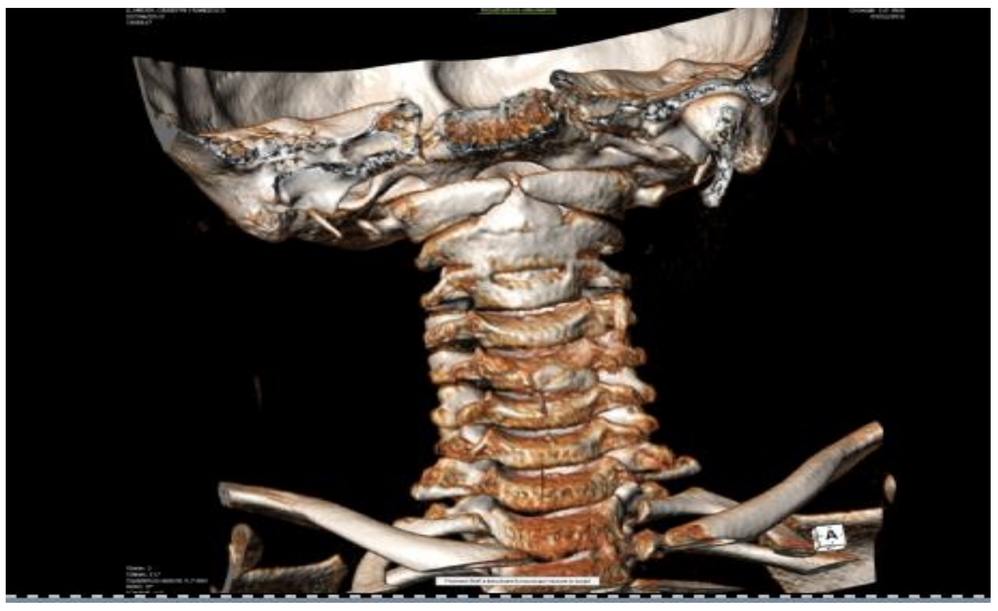
BACKGROUND

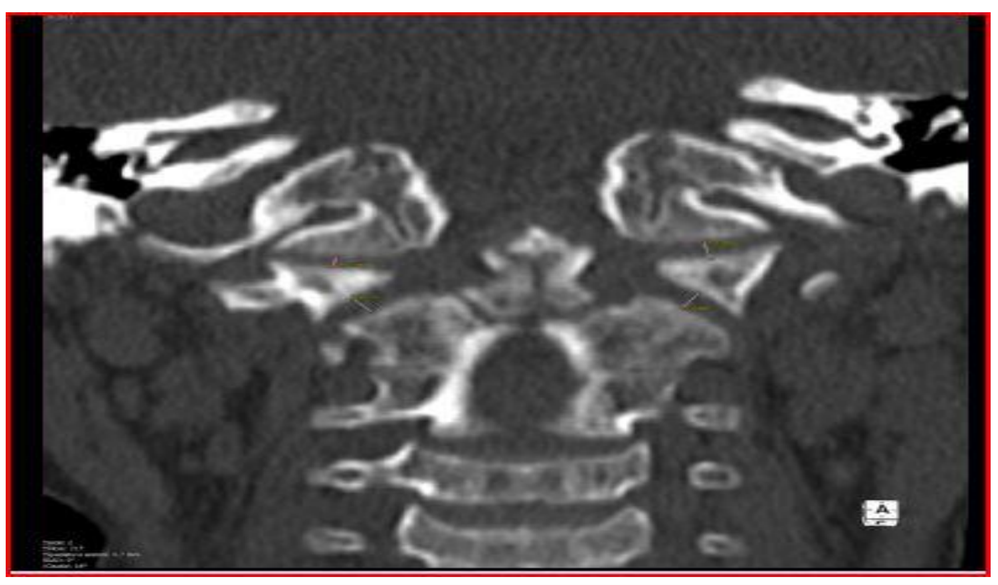
Anomalies of cervical spine have been described in association with pituitary anomalies in the context of malformative syndromes with midline defects. Several genes are involved in pituitary gland development, differentiation and function. In particular the presence of os odontoideum has been reported in a case of pituitary hypoplasia associated with leg anomalies and renal dysplasia and lack of aberrations of the BMP4, BMP2 and PTX1 genes, but duplication of pituitary gland was also reported in association with odontoid process. Here we describe a case of pituitary stalk interruption syndrome with a complex cranio-vertebral junction malformation.

CASE REPORT

A 7 year-old boy was admitted to our Endocrinology Unit for a poor growth history.
Auxological evaluation showed a severe growth delay (-3.4 DS) and delayed bone age (4 years according to the Greulich and Pyle method). Physical examination revealed dysmorphic notes (short neck, low implant ears, big and stumpy hands). Neurological evaluation showed intra-rotation of right foot, motor clumsiness, slight reduction of muscle strenght in the limbs, presence of clonus on the right foot. Because of an history of neonatal hypoglycemia and bilateral cryptorchidism, a congenital hypopituitarism was suspected. The study of pituitary function revealed a combined pituitary hormone deficiency. Therefore a substitutive therapy with rhGH, levo-thyroxine and hydrocortisone was started. The brain MRI showed evocative elements of pituitary stalk interruption syndrome PSIS (interrupted pituitary stalk, ectopic posterior pituitary, anterior pituitary hypoplasia) and, in association, complex cranio-vertebral junction anomalies that were confirmed and better defined by a 3D CT (figures) and midollar MRI. These anomalies were characterized by the following elements: presence of os odontoideum, dysmorphism of the apex of epistropheum tooth, median cleft of anterior and posterior arc of C1, synostosis of posterior arches of C2 and C3; at the site of the stenosis the cervical cord resulted compressed concentrically. The child underwent urgent neurosurgery with good post-operative course. Genetic evaluation for HESX1, LHX3, LHX4, PROP1, POU1F1, SOX3 and SOX2 is ongoing.







CONCLUSIONS

In patients with PSIS and neurological involvement, cranio-vertebral junction anomalies could be suspected and, if present, rapidly treated in order to avoid the progression towards a medullary compression. All patients with these anomalies should have genetic counseling in order to identify causative genes.