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Pallister Hall syndrome: an unusual case of central precocious puberty, prolonged vaginal bleeding, gelastic seizures and polysyndactyly in a 3 month old infant

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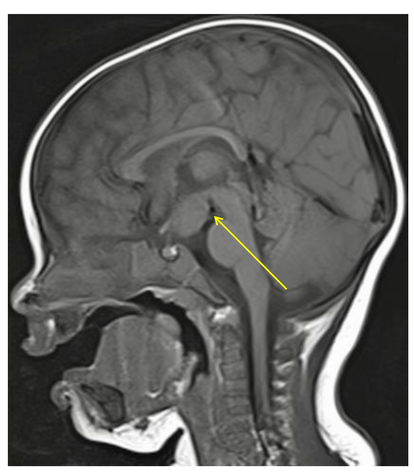
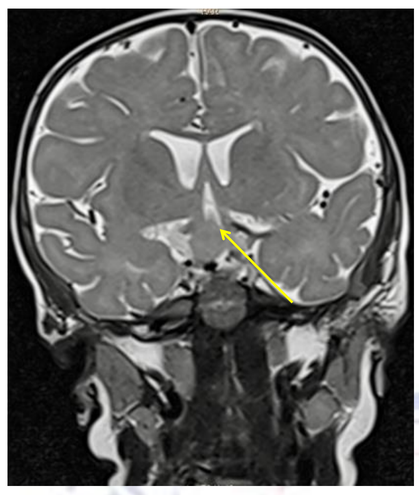
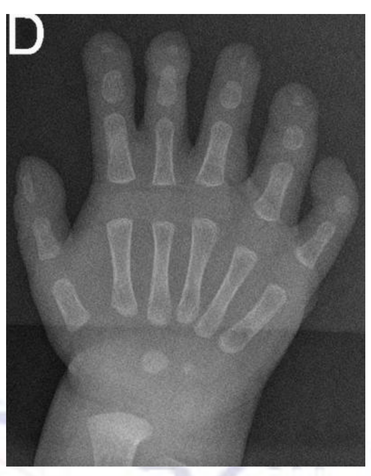
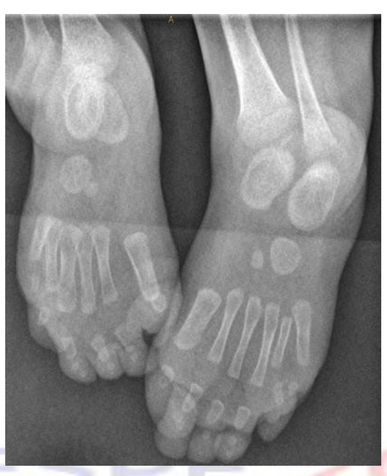
Introduction: Central precocious puberty (CPP) at a very early age is usually caused by an organic lesion. The most frequent one is the hypothalamic hamartoma (HH), which, associated with polysyndactyly, cleft palate and gelastic crises, clinically suggests the diagnosis of Pallister Hall Syndrome. Identification of a heterozyqous pathogenic variant in *GLI3* confirms the diagnosis.

Case: A 3-month-old girl with no family history consults for vaginal bleeding presented from birth, with frequencies 1-2 times a month, and bilateral breast tissue with hyperpigmented areolas.
Polydactyly in hands and feet was evident from the second trimester of pregnancy. The girl was born at 41 weeks with a weight of 3330 g, length of 52 cm, cephalic perimeter of 33 cm and a posterior cleft palate, as well as polysyndactyly in hands and feet. Echocardiography evidences ostium secundum -type interauricular communication. From the first months of life, the family reports stereotyped episodes of facial expression change, such as sudden laughing or crying of about 4-5 seconds.

Complementary test: aGnRH stimulation test (4 months of age): peak of LH 79 IU / L and FSH 16.26 IU / L, Estradiol 116.47 pg / mL
-Pelvic Pelvic ultrasound showed the uterus increased in size, with a cervix / fundus ratio greater than 1/1 with marked endometrial thickening.
-MRI showed an oval, solid, suprasellar lesion measuring 20 x 18 x 13 mm in diameter, compatible with pedunculated hypothalamic hamartoma.
-Genetic study of *GLI3* gene: pathogenic mutation not described: exon 15: c.3439G> T confirming the diagnosis of Pallister Hall syndrome.

Evolution and treatment: The patient was treated successfully with GnRh analogues (aGnRH), with an initial dose of 1,87 mg at 0, 14 and 28 days and then with 1,87 mg every 28 days. Gelastic seizures were treated with oxacarbamazepine.



Conclusion: CPP at an early age with prolonged vaginal bleeding may be caused by an organic cause. Phenotypic anomalies with gelastic seizures lead us to a Pallister Hall syndrome. Our patient is one of the youngest infants presenting with CPP and HH in all of Europe. The treatment was successful after the first doses of aGnRH