PRECOCIOUS PUBERTY IN SEPTO-OPTIC DYSPLASIA SYNDROME PRESENTATION OF 2 CASES

A. Malinowska [1], E. Marczak [1], A. Rudzka-Kocjan [1], M. Szalecki [1,2]

1.Department of Endocrinology and Diabetology, Children's Memorial Health Institute (CMHI), Warsaw, Poland
2. Faculty of Medicine and Health Sciences, University of Jan Kochanowski (UJK), Kielce, Poland

Introduction

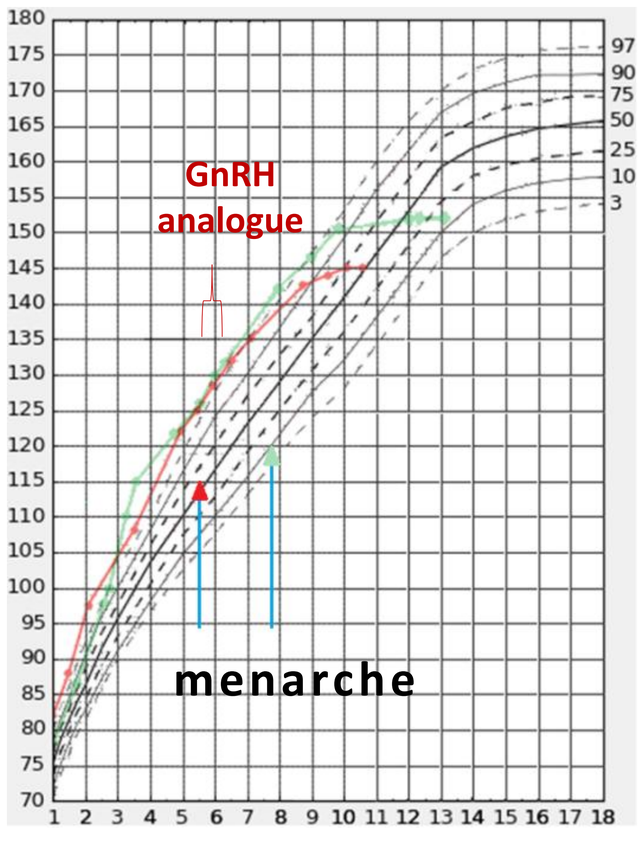
Septo-optic dysplasia (SOD) is a rare, congenital condition that can be caused by mutations in HESX1, OTX2, SOX2, SOX3 genes. Symptoms of SOD include: optic nerve hypoplasia, hypopituitarism and midline brain abnormalities such as absence of septum pellucidum and/or corpus callosum. Hypopituitarism in SOD usually manifests as growth hormone deficiency followed by central hypothyroidism as well as deficiency of gonadotropins. Precocious puberty is a rare finding among patients with SOD, though it appeared among 2 of our patients.

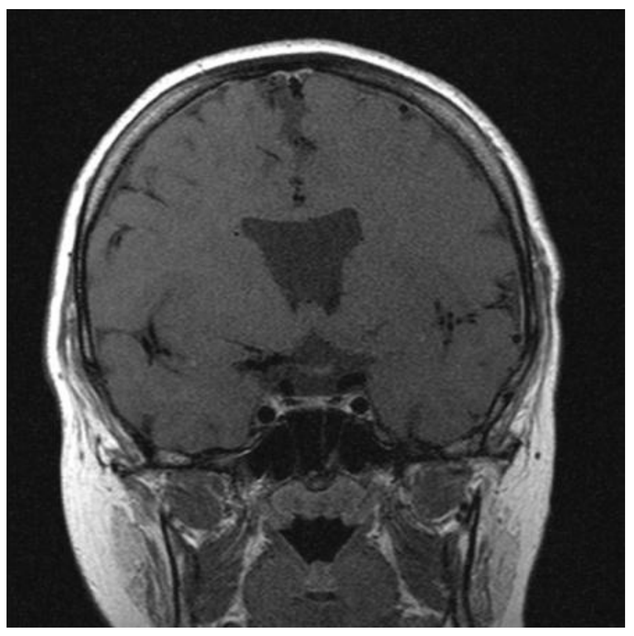
Materials

2 girls diagnosed with SOD at the age: **Patient No 1** - 3rd year of life and **Patient No 2** – first year of life.
At the moment girls are 10,5 and 13 years old.

Both patients had:
- hypernatremia observed since neonatal period
- Diagnosis: diabetes insipidus
  TREATMENT → **DESMOPRESSIN**
- blindness caused by bilateral optic nerve hypoplasia
- developmental delay
- lack of septum pellucidum (MRI)
- obesity, tall stature and since 1st year
- accelerated bone age
- TSH insufficiency TREATMENT → **L-THYROXINE**
- GH deficiency
- ACTH insufficiency excluded

Patients did not receive:
- Growth Hormone treatment
- Hydrocortisonum



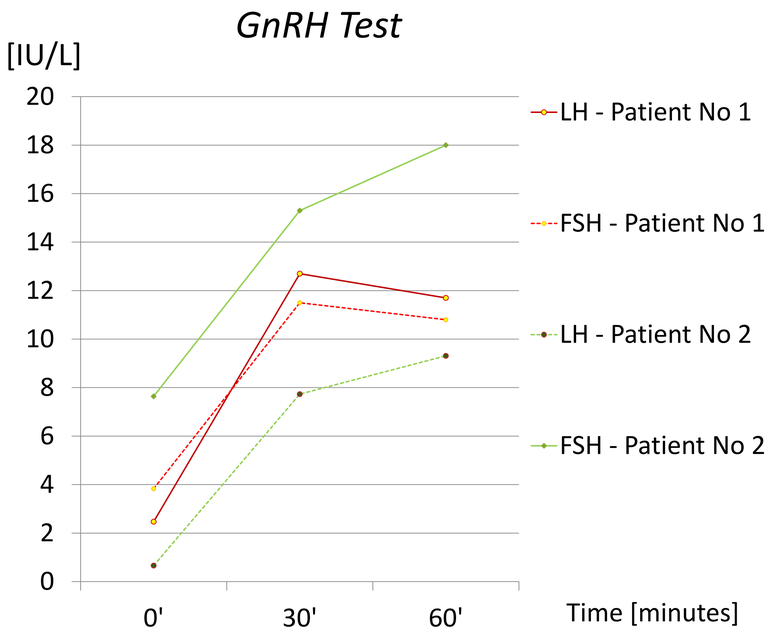


Puberty signs:
- thelarche: 5 yrs
- menarche: 5,5 and 8 yrs
- accelerated bone age: 3 yrs and 2,6 yrs
- Labs: pubertal levels of gonadotropins in GnRH test, high levels of estradiol

Diagnosis: central precocious puberty

Patient No 1 received TREATMENT → **LONG-ACTING GnRH ANALOGUE** because of rapid progression of puberty.
Medication was withdrawn within less than 6 months due to recurrent urticaria.
Patient No 2 was not treated with GnRH analogue.

Results



Hormone	Patient No 1	Patient No 2	Ranges
IGF-1 [ng/mL]	21	37	82-262
E2 [pg/mL]	31,9	77,6	<8
PRL [ng/mL]	7,04	9,77	2,7 – 19,7
TSH [ng/dl]	0,24	0,03	0,4-6,0
fT4 [pmol/L]	0,78	0,72	1,8-4,1
Cortisol – baseline [ug/dl]	5,2	3,3	5-20
Cortisol in 60' ACTH stimulation test [ug/dl]	26,6	24,2	

Conclusions

In most cases SOD is associated with multiple pituitary hormone deficiency, including gonadotropins. This report implicates the necessity for long-term monitoring as pituitary insufficiency may evolve over time and does not rule out precocious puberty.

References

1. De Morsier G. Etudes sur le dystrophies cranioencephaliques III. Agenesie du septum lucidum avec malformation du tractus optique. Neurochirurgie et Psychiatrie1956.
2 Hoyt WF, Kaplan SL, Grumbach MM & Glaser JS. Septo-optic dysplasia and pituitary dwarfism. Lancet 1970.
3 Stanhope R, Preece MA & Brook CGD. Hypoplastic optic nerves and pituitary dysfunction. a spectrum of anatomical and endocrine abnormalities. Archives of Disease in Childhood 1984.

55ESPE 742--P1
Pituitary and Neuroendocrinology
Anna Malinowska
DOI: 10.3252/pso.eu.55ESPE.2016
Poster presented at: ESPE 2016 PARIS
PosterSessionOnline.com