Congenital panhipopituitarism and ectopic posterior pituitary

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Introduction

Congenital hypopituitarism may be the result of complications linked with delivery or may sometimes result of insufficient development of the gland in the context of specific genetic abnormalities(PROK2 and PROKR2, LHX4, HESX1, OTX2, GLI2 and SOX3). Interruption or lack of pituitary stalk represents a frequent feature of congenital hypopituitarism.

Case description

We present a patient 39 years old with congenital panhypopituitarism, mental retardation, dysmorphic syndrome and diabetes type 1 but without diabetes insipidus.
MRI imaging showed ectopic posterior pituitary at the tuber cinereum level associated with the lack of pituitary stalk.
Medical management consists of replacement therapy for all lines deficient hormones and insulin therapy.

Results

Laboratory findings – baseline tests - have demonstrated combined multiple pituitary hormone deficiency involving growth hormone, thyrotropin, adrenocorticotropic and gonadotropics hormones without response to dynamics tests (insulin tolerance test and releasing factors TRH, GnRH and CRH).

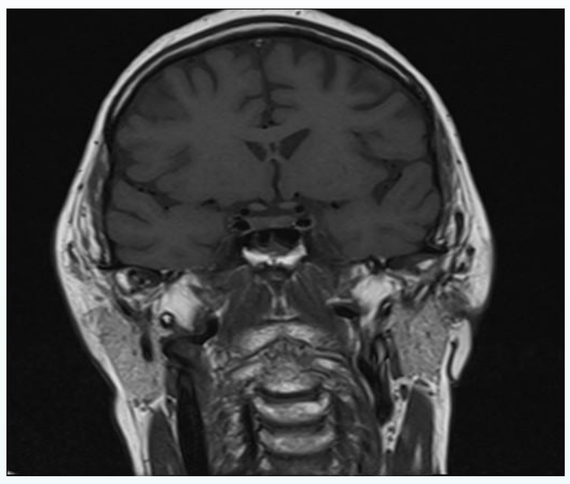
Baseline tests

GH	<0.5 ng/ml
IGF1	35 ng/ml
TSH	1.41 mU/l
fT4	0.4 ng/dl
fT3	1.2 pg/ml
ACTH	12.2 pg/ml
Cortisol	2.1 µg/dl
LH	<0.2 mU/ml
FSH	<1 mU/ml
Free Testosteron	0.28 ng/dl

Dynamics tests

Test		0’	30’	60’	90’	120’
With GHRH	GH (ng/ml)	<0.5	<0.5	<0.5	<0.5	<0.5 l
With TRH	TSH (mU/l)	0.03	0.15	0.14	0.11	0.10
With CRH	ACTH (pg/ml)	18.9	17.8	18.8	16.6	18.9
	Cortisol (µg/dl)	2.2	2.2	1.9	1.4	1
With GnRH	LH (mU/ml)	<0.2	<0.2	<0.2	<0.2	<0.2
	FSH (mU/ml)	0.2	0.3	0.2	0.4	0.5

MRI pituitary showed ectopic posterior pituitary at the tuber cinereum level associated with the lack of pituitary stalk.



MRI pituitary

DXA

BMD lumbar spine (L2-L4)= 0,641 g/cm2,
Z- score= -3,8DS , T-score= -4,1DS.
BMD right hip = 0,432 g/cm2,
Z- score= -3,7DS , T-score= -4,0 DS.

Treatment
Medical management consists of replacement therapy for all lines deficient hormones and insulin therapy.

Conclusion

The patients with no visible pituitary stalk on MRI present a more severe form of the disease associated with multiple deficiency of the anterior pituitary hormones, whereas the presence of the pituitary stalk leads to isolated GH deficiency. Follow-up on these patients is necessary, as the natural history of the disease is not established until adulthood.

Key words

Congenital hypopituitarism , pituitary stalk , ectopic posterior pituitary