Septo-optic dysplasia and excellent growth with low Growth Hormone dose: our experience

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OBJECTIVES

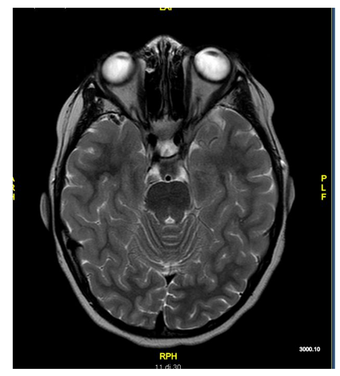
Septo-optic dysplasia (SOD) is a congenital, rare anomaly (1:10000) associated with optic nerve hypoplasia, midline brain abnormalities and pituitary hormone abnormalities (1). Genetic alterations associated are: HESX1, SOX2 and SOX3. SOD's pattern of growth even without GH treatment has been reported to be often good (2,3).

METHODS

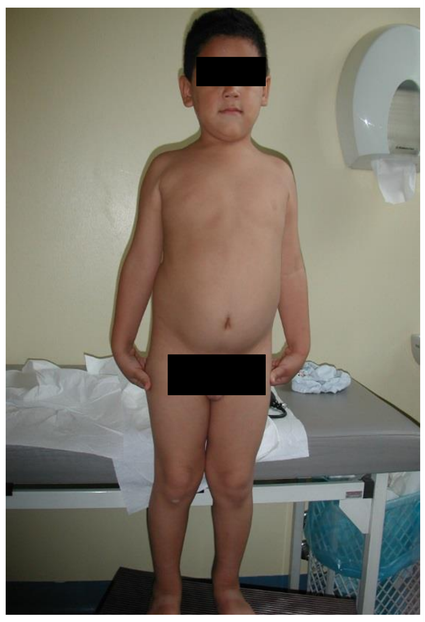
We present our experience

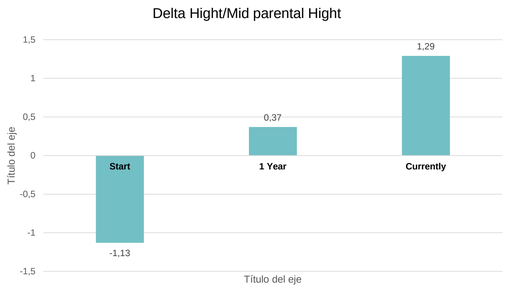
Number	5	
M:F	4:1	
BW (SDS)	-0.25	
BL (SDS)	0.23	
Hormone Deficiency	GH	100%
	TSH	100%
	ACTH	80%
	Gonadotropin	40%
	Central Diabetes Insipidus	20%
MRI	Ectopic neurohypophysis	100%
	Optic nerve hypoplasia	100%
	Chiasm hypoplasia	100%
	Hypophysis Hypoplasia	80%
	Stalk hypoplasia	80%
	Stalk absent	20%

Genetic	HESX1 +	20%
Start of therapy	Age	1.38ys
	Height (SDS)	-1.32
	ΔMph (SDS)	-1.13
Currently	Age	11.8ys
	Height (SDS)	1.09
	ΔMph (SDS)	1.29



Years of treatment	9.83ys		
Height gain (SDS)	1st year	+1.34	
	Total	+2.54	
Growth velocity (SDS)	1st year	+1.97	
	Currently	+1.02	
ΔMph	Start	-1.13	
	1st year	0.37	
	Currently	1.29	
IGF-1 (SDS)	Start	<-2	<u>≤25</u> (55-327)
	Least once	>+2	Pt1 <u>651</u> (57-316)
			Pt2 <u>462</u> (74-388)
			Pt3 <u>364</u> (52-297)
			Pt4 <u>430</u> (74-388)
			Pt5 <u>266</u> (79-244)
Dose (mg/kg/wk)	Start	0.17	
	Currently	0.11	
	Total	0.18	





RESULTS

All our patients presented as starting symtoms severe GH deficiency with low IGF1 levels, hypoglycemia and pathological MRI, therefore started GH treatment before 3 ys of age. During GH treatment growth rate was excellent with a starting GH dose of 0,18 mg/kg/wk. As IGF1 levels were >+2 SDS in each patients at least once, GH posology was reduced to a median of 0,11 mg/kg/wk with a normalization of IGF1 levels, nevertheless growth velocity remained good (gv: +1,02 SDS), with a currently median height gain of +2.54 SDS and a median delta to midparental height of +1.29 SDS after almost 10 years of treatment.

60% (3/5) of our patients present excess of weight (2 overweight, 1 obese); with no evidence of hyperinsulinemia.

CONCLUSIONS

Five SOD patients followed in our Centre, with GH and IGF1 deficiency, present excellent growth during GH treatment even with minimal doses, as previously reported in the literature. We confirm the known tendency to present excess of weight (3), with no evidence, in our patients, of hyperinsulinemia. The mechanism of this excellent growth is not clear: high levels of insulin, PRL or leptin may be involved; the trend to overweight may partially explain this phenomenon. Actually no specific cause has been found, therefore more studies are needed to better explain this phenomenon.

References

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Growth

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