IGFALS Gene Deletion in a Family with Short Stature

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Backround:

ALS deficiency is characterized by mild short stature, delayed puberty, low serum IGF1, low serum IGFBP3 and undetectable serum ALS levels.

Case:

11.3 years old, boy, presented with short stature.

Term, Birth weight ? (unknown), consanguineous parents

Height: 130.5 cm (-2.33 SDS)

Weight: 25.2 kg (-2.36 SDS)

BMI: 25.2 kg (-1.6 SDS), HC: 49.8 cm

US/LS: 1 (+1>-<0 SDS), AS-Height: -5.5 cm (-1>-<-2 SDS)

System exam were normal, TV 2/2 cc, Pb2

CA: 11 y 3/12 m,

BA: 9 y

PAH: 165.6 cm (-1.68 SDS)

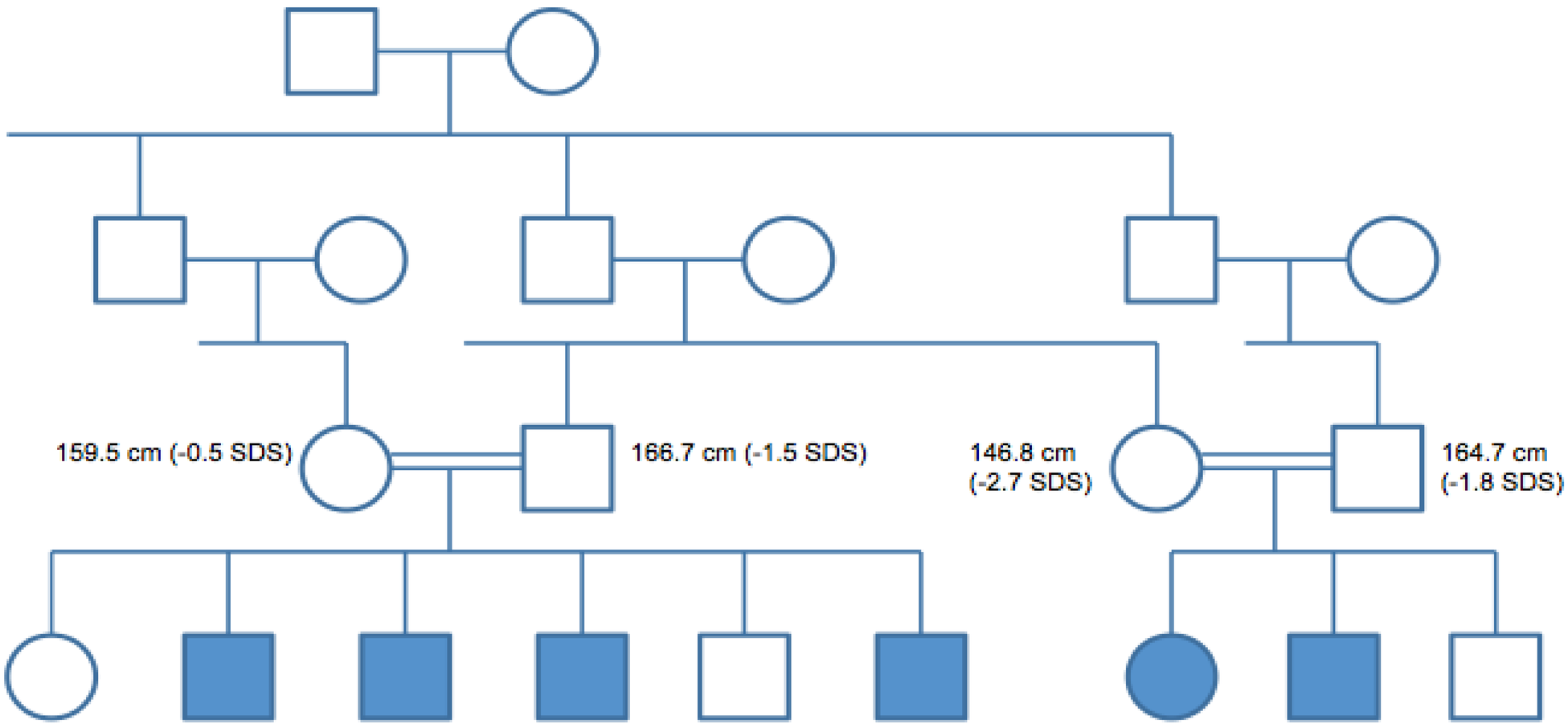
MPH: 169.6 cm (-0.99 SDS)

IGF-1: 37.6 ng/ml (111-551 ng/ml)

L-dopa peak GH: 10.3

Table 1: IGF generation test

	Before	After
IGF-1 (ng/ml)	37	39
IGFBP3 (ng/ml)	<500	<500



	1	2	3 (index)	4	5	6		7	8	9
Age	15.5	13.7	11.3	9	6.8	4.3		9.5	7.1	4.3
Height SDS	-0.87	-2.36	-2.33	-1.76	-1.33	-2.02		-2.93	-3.64	-1.60
Weight SDS	-1.65	-1.78	-2.36	-2.16	-1.53	-2.84		-2.41	-4	-1.1
IGF-1 (ng/ml)	-	41	37	25	-	40		29	<25	51
IGFBP3 (ng/ml)	-	<500	<500	<500	-	1470		<500	<500	1300
ALS (mg/L) (SDS)	13.0 (-1.5 SDS)	1.50 (-3.8 SDS)	1.54 (-3.49 SDS)	1.50 (-3.28 SDS)	10.8 (-0.71 SDS)	5.56 (-2.0 SDS)		1.44 (-4.3 SDS)	1.50 (-3.0 SDS)	4.52 (-2.3 SDS)
IGFALS gene (c.1477del)	+/-	-/-	-/-	-/-	+/+	+/-		-/-	-/-	+/-

Conclusion: Since, patients with ALS deficiency have mild short stature and heterozygous parents have low-normal height, these patients can be diagnosed as familial short stature. Therefore, the physician should be careful at differential diagnosis of short stature.

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