Short stature in Protein Arginine Methyltransferase 7 (PRMT7) Mutations: first evidences of growth response to GH treatment

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INTRODUCTION

Protein arginine methyltransferase 7 (PRMT7) is a member of a family of enzymes that catalyzes the transfer of methyl groups from S-adenosyl-L-methionine to nitrogen atoms on arginine residues involved in multiple biological processes, such as signal transduction, mRNA splicing, transcriptional control, DNA repair and protein translocation.

Currently, 12 patients with homozygous/compound heterozygous mutations in PRMT7 gene have been described defining the human disorder known as SBIDDS syndrome (OMIM #617157).

CASE REPORTS

Due to severe short stature and growth impairment endocrine investigations to rule out GHD were performed:

Twin A:
- Peak GH at arginine test: 4.61 µg/L;
- Peak GH at glucagon test: 5.14 µg/L;
- IGFI: 47 ng/mL; -2.19 SDS.

MNI:
- Normal hypothalamic-pituitary region
- Corpus callosum thickening

Twin B:
- Peak GH at arginine test: 11.8 µg/L;
- IGFI: 52 ng/mL; -2.05 SDS.

MNI:
- Dysmorphic features of corpus callosum
- Corneal folds
- Pars intermedia cyst

GHD 0.025 mg/kg/day

CONCLUSIONS

Our findings provide further clinical data and expand the knowledge of endocrine manifestations associated with PRMT7 homozygote/compound heterozygote mutations including GHD.

Considering short-term good response to rGH, further studies are needed to confirm long-term outcomes at adult height and establish whether SBIDDS could be considered, among those syndromes treatable with rGH.

REFERENCES


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