

Title: GH deficiency and growth retardation in Rett syndrome

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Text body: Growth retardation is one of the principal features of Rett Syndrome, being one of diagnostic criteria and a specific outcome marker. Standard growth charts don't allow clinicians to differentiate growth trends of Rett patients from the ones of other individuals affected by other cause of growth retardation (such as malnutrition or other diseases), by the way specific curves for this syndrome have never been validated.

Even if malnutrition seems a reasonable cause of growth retardation in these individuals, in a relevant amount of them growth retardation raises up with a balanced diet and an adequate BMI. Whether an alteration of the Ghrelin-GH-IGF1 axis could be the cause of this problem is controversial; to our knowledge there are no data reporting a GH deficiency in Rett patients.

In this report we review the literature about the possible correlation between Ghrelin-GH-IGF1 axis alteration and growth delay in Rett syndrome, describing the case of a female patient affected by the classical variant (MECP2 R168X mutation in heterozygosis) who started showing signs of growth retardation, consequently targeted as a consequence of pituitary dwarfism.

Investigations on pathogenetic triggers and mechanisms that carry to growth retardation in Rett Syndrome are needed to meliorate therapy and outcome of patients affected by Rett Syndrome.

References

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