Growth Hormone Excess in McCune-Albright Syndrome

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BACKGROUND AND OBJECTIVES

McCune-Albright Syndrome is a combination of polyostotic fibrous dysplasia (BFD), café-au-lait skin pigmentation and hyperfunctioning endocrinopathies. It results from postzygotic mutations in a subunit of the Gsalfa protein and the consequent phenotype is a mosaic with high degree of clinical variability. The aim of the study is determine prevalence and characteristics of GH hypersecretion (GHH) in MAS.

METHODS

We screen GH excess in two cohort of MAS patients: 3/34 cases (92%) and 28/129 cases (21.7%). Average age at diagnosis is 13.9 y.o. (from 3 to 36). Male are 18/31 (58%), female are 13/31 (42%), age range at last evaluation 5-49 y.o. (mean age 21.1 y.o.). In all we study auxological data, biochemical GHG (IGF1 Z score, random GH, GH after OGTT), association with prolactin hypersecretion (PH), possible abnormal pituitary MRI, BFD, response to medical and other treatment. 30 MAS cases with GH excess were matched with 30 MAS controls without GH excess for sex, age (± 7 y.o.) and total bone scan score at Technetium Tc 99m bone scintigraph (± 3 units) to evaluate association between comorbidities and GH hypersecretion. At the end we divided patients with GH excess in two groups: Group A (17 MAS, therapy before 20 y.o.) and Group B (13 MAS, no therapy or therapy after 20 y.o.).

RESULTS

PH occurs in 27/31 (87%). Pituitary adenoma is evidenced in 16/31 (52%), while craniofacial and long bone FD are evidenced in 100%. 23/31 have also Hyperprolactinemia (74.1%). Medical treatment was performed in 25/31: in 17 ocreotide 10-30 mg im/month, in 5 ocreotide 30 mg im/month, and pegvisomant 20 mg s.c./day, in 1 ocreotide 30 mg i.m./month, pegvisomant 20 mg s.c./day and pituitary irradiation, in 2 ocreotide 30 mg i.m./month and transphenoidal pituitary surgery (1 died for post-operative complications). 19/24 (79%) have complete control of GHH (Z < IGF1 Z-score < +2), 3 patients are non-compliant to therapy.

CONCLUSIONS

GH excess is present in about 20% of MAS patients, 76% of them have also Hyperprolactinemia. MAS phenotype with GHH is more severe because always associated with cranio-facial FD, head circumference expansion and more comorbidities (facial asymmetry, optic neuropathy and hearing deficit). Pain is not affected by GHH Hypersecretion. Early therapy should be effective in preventing Optic Neuropathy.

References
