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Familial Carney Complex: Follow Up of a Family With Negative PRKAR1A Genetic Screening

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Background: Carney’s complex (CNC) is a rare genetic multiple neoplasia syndrome, with involvement in several systems, among them, acromegaly is a possible condition, according to the literature, seen in about 10% of cases. PRKAR1A is the most known gene evolved. The genetic origin of 59% of patients with CNC has not yet been elucidated. Clinical Cases: A 17-year-old male with tall stature, with stigma of acromegaly, GH 9.89 ng/ml and IGF-1 1.5 times higher reference value, MRI with pituitary adenoma of 0.7cm. After resection transfenoidal endoscopic (TSE) the pathology of the tumor revealed a pure GH pituitary adenoma. Echocardiogram showed cardiac myxomas. For that reason, a Carney complex was established. In the search for other alterations present in the syndrome, a nodule was found in the right testicle, which after nodulectomy shown a Leydig Cell Tumor. After these findings, we invited all family members to perform screening to identify possible involvement of the CNC. Two brothers, including a twin, and their mother also had stigma of acromegaly, all with pituitary adenoma and laboratorial diagnostic. The two men underwent TSE surgery reassuring GH secretion by the tumor. The elderly brother had aggressive microcalcifications in bilateral testicles. After bilateral orchiectomy was diagnosed with large cell calcifying sertoli cell tumor. The twin brother also had cardiac mixomas and macrocalcifications in testicle but without signs of malignancy. The mother had athyroid ultrasound showing a multinodular goiter and breast lesions also with no signs of malignancy. She also had a pituitary microadenoma, but refused the surgery and is currently using a somatostatin analog. CNC is a multiple neoplasia syndrome usually characterized by lentiginosis, multiple neoplasias and signs of endocrine overactivity, particularly Primary Pigmented Nodular Adrenal Disease (PPNAD) and myxomas. It is a rare condition and its prevalence remains unknown. A patogenic allelic variant (PAV) of PRKAR1A is found in 37% of patients with sporadic CNC and more than 70% of patients with familial CNC. To this date, at least 130 different mutations have been described in more than 400 families around the world, distributed across the ten coding exons (PRKAR1A has 11 exons, but exon 1 is non-coding and rarely mutated) and in the adjacent intronic sequences. In 2014, a germline triplication of the exon of PRKACB gene has been identified in a patient who did not harbour a PRKAR1A mutation. In our case, a search of PAV was realized by exome NGS and CG-Array but we found no evidence of variant that can explain the occurrence of CNC.

Conclusion: We describe the follow-up of a family with CNC without PAV identified so far.

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