A microdeletion of *PRKARIA* associated with Carney Complex

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Introduction

Carney Complex (CNC) is a rare multiple neoplasia syndrome, its commonest endocrine manifestation being ACTH-independent Cushing's syndrome, that is histologically characterized by primary pigmented nodular adrenocortical disease (PPNAD) (1).

There is significant genetic and phenotypic heterogeneity, but deletions at 17q24.2 are rare (2).

We describe the particular characteristics of a patient with a microdeletion in this area.

Case report

A 37-year-old male was referred to the Endocrine Consult Service by the Internal Medicine department, following an episode of severe hyponatremia with altered consciousness and rabdomyolysis.

He had a history of cyclical Cushing's syndrome from the age of 5 and had bilateral adrenalectomy at the age of 10 (3). He had been on lifelong hydrocortisone and fludrocortisone replacement, but was not adherent to the medications, due to concurrent problems with alcohol abuse.

His GH, IGF-1 and prolactin measurement were normal

He had suffered a non traumatic subcephalic right femoral neck fracture two years previously and had a total hip BMD of 0.604 and a Z-score -2.8 on the left.

On examination, he was lean, with pectus excavatum, thoracic spine scoliosis, small testes (8 and 10 ml), and multiple lentigines on the trunk and buccal mucosa. (Figure 1).

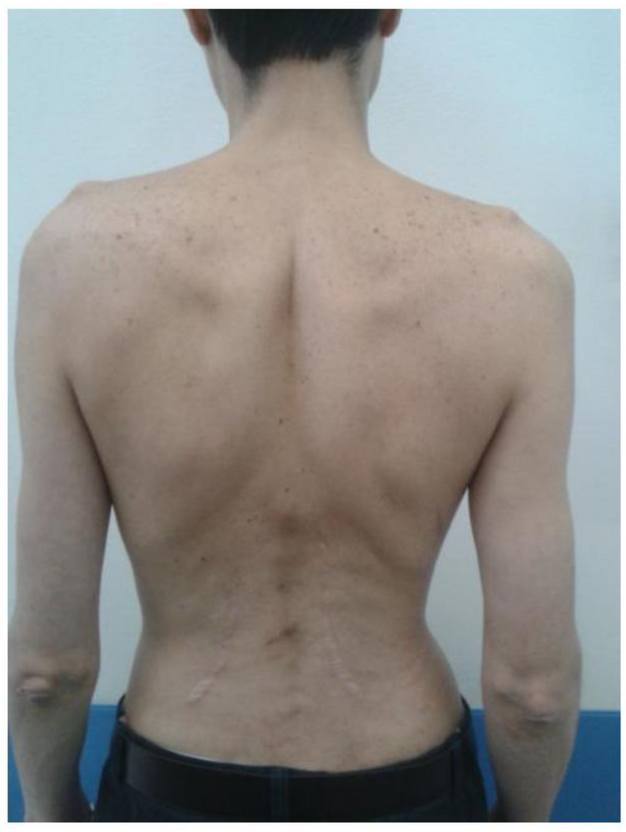

Fig.1

The patient had a normal echocardiogram, testicular ultrasound was significant for bilateral micro calcifications and he had no other clinical or laboratory evidence of endocrine dysfunction.

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Chromosomal microarray analysis revealed a 0.98 kb deletion at 17q24.2.

Testing of his mother and sister detected no genomic imbalance of 17q24.2. Because the patient's father was deceased, it is not feasible to ascertain whether this was a *de novo* or inherited mutation.

Conclusion

Despite significant but also overlapping phenotypic and genetic heterogeneity, PPNAD-whether clinically indolent or apparent- is the most frequent endocrine manifestation of CNC and should prompt genetic confirmation and long term surveillance for other syndromic manifestations.

References

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