Incidental pituitary adenoma detection in two patients affected by Williams syndrome: only a coincidence?

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INTRODUCTION

Williams Syndrome (WS) is a multisystem disorder caused by a deletion of part of chromosome 7 (del7q11.23).

The birth prevalence is 1:7500. M:F=1:1.

CASE REPORTS

Patient 1, female

WS genetically diagnosed at 11.8 years of age based on typical facial features, mental retardation (IQ 34) and chronic constipation.

Pregnancy and neonatal period unremarkable. No cardiac defects. Satisfying growth.

Cerebral MRI (performed during the diagnostic work-up for neurodevelopmental impairment at 11.25 years) showed enlarged pituitary (height of 9 mm) in the context of which a mass with suprasellar extension was detected.

Patient 2, female

WS genetically diagnosed at 20 months of age based on failure to thrive, typical facial features and mild neurodevelopmental retardation.

Born small for gestational age (SGA). No cardiac defects. At the age of 9.5 years, diagnosis of growth hormone deficiency.

Cerebral MRI (performed at 10 years of age as part of the diagnostic work-up of GHD) showed a lesion 5 mm large sited at the anterior side of the pituitary stalk at an intra-suprasellar level.

CONCLUSIONS

Blood test | Patient 1 | Patient 2
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GH (0.01-6.20 ng/ml) | - | 9.71
IGF-1 (49-504 ng/ml) | 375.5 | 392.7
TSH (0.35-4.94 mIU/ml) | 4.02 | 4.98
PRL (3-27 ng/ml) | 16 | 20.9
LH | 4.2 | 1.6
FSH | 5.6 | 5.2
Calcium (8.5-10.5 mg/dl) | 9.2 | 9.5

Both patients did not complain of any visual problems nor headache.

Up to today, only radiological, biochemical and clinical follow-up has been indicated for both girls.

REFERENCES


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