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

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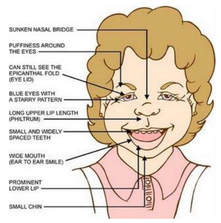
INTRODUCTION

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



Main features
Dysmorphic facies (100%)
Cardiovascular disease (80%; most commonly supravalvular aortic stenosis)
Intellectual disability (75%)
Characteristic cognitive profile (90%; es cockatil party personality)
Idiopathic hypercalcemia (15% to 45%)

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 neurodevelopment impairment at 11.25 years) showed enlarged pituitary (height of 9 mm) in the contest 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.

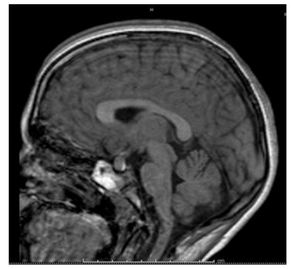
Born small for gestational age (SGA). No cardiac defects. At the age of 9.5 years, diagnosis of growth hormone deficiency.

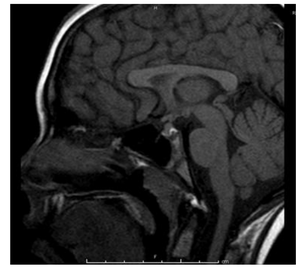
Cerebral MRI (performed at 10 years of age as part of the diagnostic work-up of GHD) showed a lesion 5 mm large sited at the anterior side of the pituitary stalk at an intra-suprasellar level.

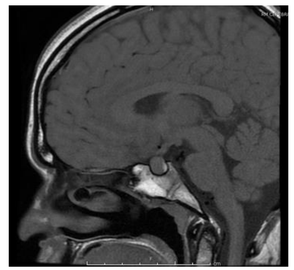
Blood test	Patient 1 1°	Patient 1 Last	Patient 2 1°	Patient 2 Last
GH (0.01-6.20 ng/ml)	-	9.71	2.48	-
IGF-1 (49-504 ng/ml)	375.5	392.7	214.2	379.6
TSH (0.35-4.94 mIU/ml)	4.02	4.98	4.44	3.83
PRL (3-27 ng/ml)	16	20.9	14.4	-
LH	4.2	1.6	1.3	5.9
FSH	5.6	5.2	8.2	9
Calcium (8.5-10.5 mg/dl)	9.2	9.5	9.4	9.3

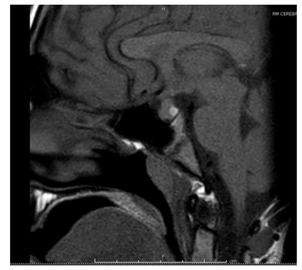
Both patients did not complain of any visual problems nor headache.

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









Patient 1: *at the top first MRI, PA max diameter 9 mm.*
At the bottom last MRI (after 2.2 years), PA max diameter 10 mm, minimal compression of the optic chiasm.

Patient 2: *at the top first MRI, PA max diameter 5mm.*
At the bottom last MRI (after 2.5 years), PA max diameter 10.5 mm, extension to the optic chiasm without compression.

DISCUSSION

- Pituitary adenomas (PAs) represent approximately 3% of all diagnosed intracranial tumors in childhood.
- 3.5-8.5% of all PAs are diagnosed before the age of 20 years, mainly during adolescence.
- Among PAs requiring treatment, only 2-6% occurs in children.
- Microadenoma >macroadenoma; secreting adenomas > non secreting-adenomas.
- PAs represent an incidental finding in 0.2% of children undergoing brain imaging.
- Growth delay could be an early symptom and should be promptly detected.
- PAs are described in some familial syndromes caused by germline genetic defects; also some somatic genetic defects have been described associated with PAs. To the best of our knowledge, PAs have not been described yet in association with Williams syndrome.

CONCLUSIONS

Growth delay, visual disturbances, headache or endocrine disorders could suggest PAs diagnosis.

Further investigations are needed to understand the possibility of a correlation between PAs and WS.

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