A larger deletion of the Williams Beuren Syndrome region encompassing genes implicated in Autism Spectrum Disorder: Case report

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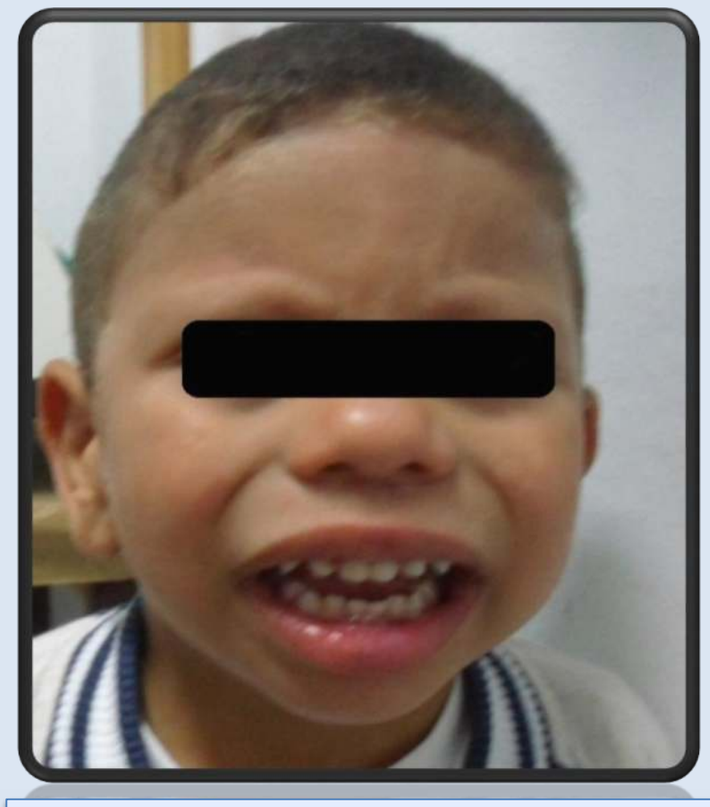
I) INTRODUCTION :

Williams Beuren Syndrome (WBS): Well-studied multisystemic disorder caused by a 1.5Mb hemizygous deletion of about 28 genes on **7q11.23**.

Characteristic pattern of symptoms: typical facial dysmorphisms, congenital cardiopathy, weakness of connective tissue, intellectual disability and a **characteristic cognitive profile** that includes relative **strengths in verbal term memory and lexical comprehension**.

II) MATERIELS ET METHODES :

❖ **4 year-old boy**



❖ **Phenotype:**

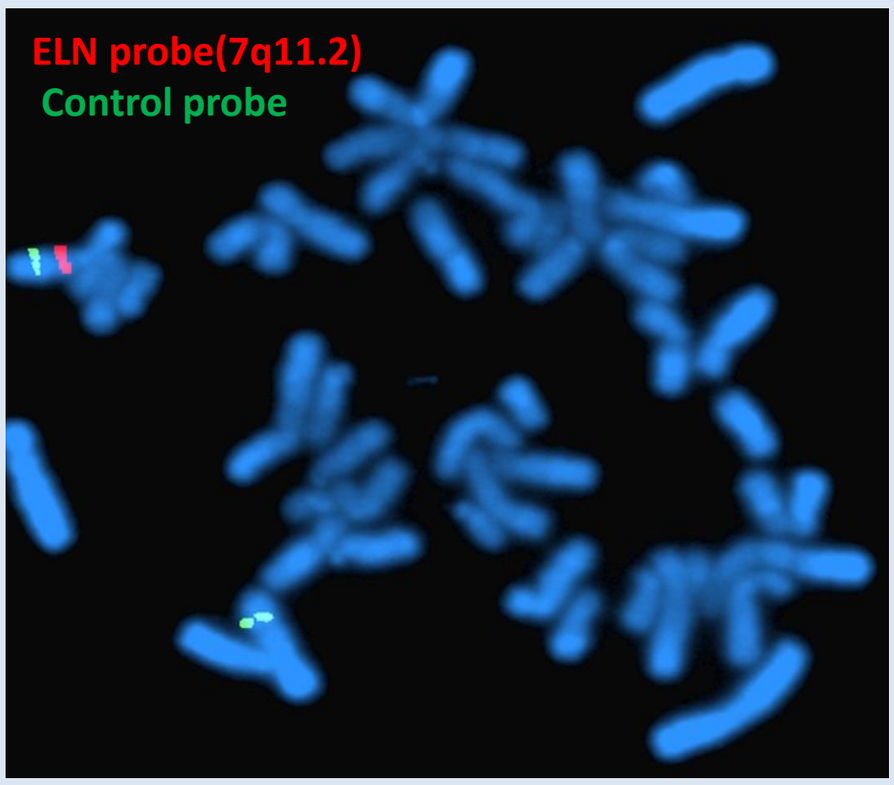
- Typical facial of WBS
- Pulmonary valve stenosis
- Development delay
- Autism Spectrum Disorder (DSMV)
- Absent language

❖ **Cytogenetic explorations:**

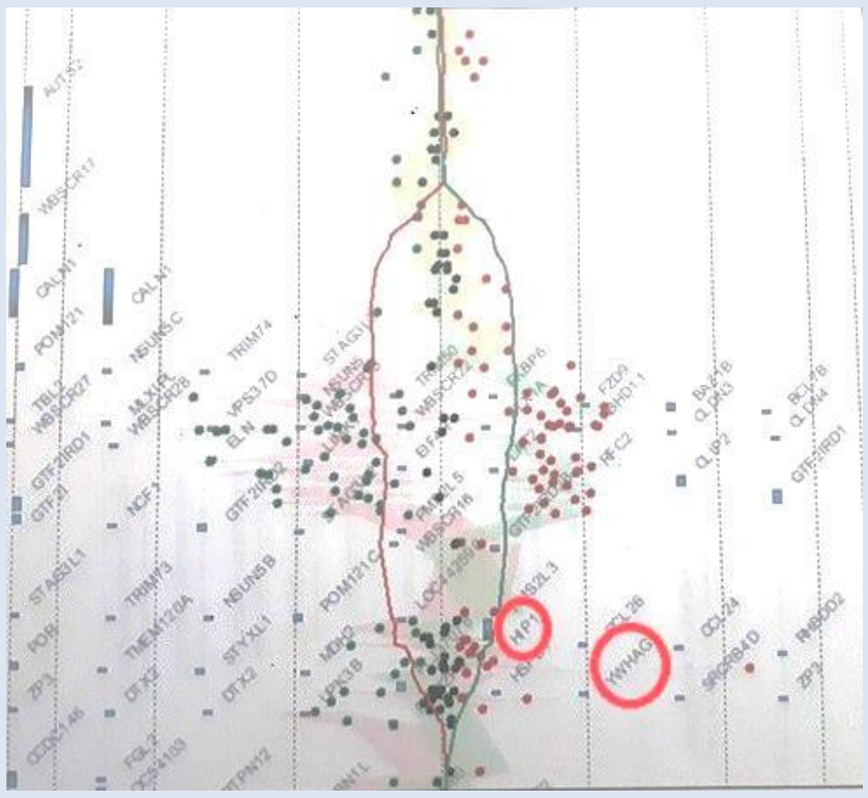
- Karyotype/ Fluorescent in situ hybridization (**FISH**)
- Array Comparative Genomic Hybridization: **aCGH** (Agilent Human Genome Microarray 44K)

III) RESULTS :

- **Karyotype:** 46,XY



- *FISH: 46,XY.ish del (7q11.23) (ELN*1)*



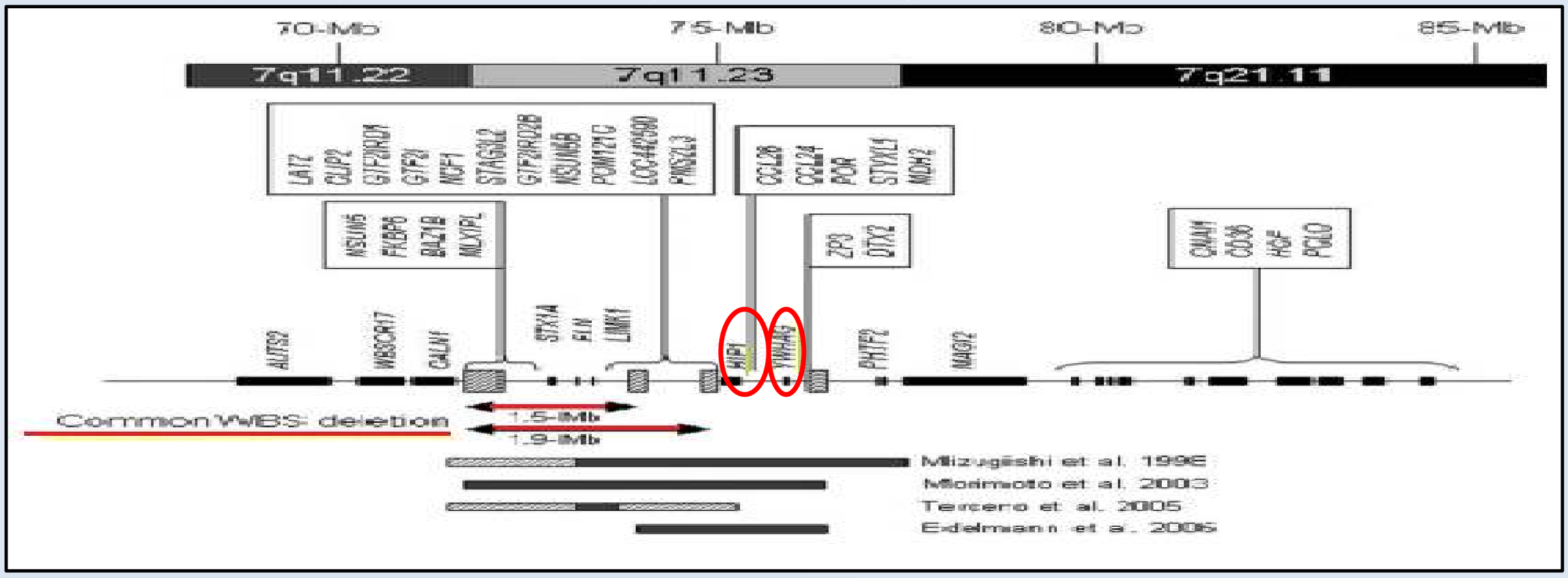
- aCGHarray: **7q11.2 deletion**

→ A larger atypical de novo (3.17Mb) WBS deletion

IV) DISCUSSION:

- WBS patients with uncommon deletion size are of particular interest for genotype-phenotype correlation.
- A **larger WBS deletions** show a more severe phenotype with serious impairments in cognitive function.

→ More severe phenotype with serious impairments in cognitive function



HIP1, YWHAG

- Deleted distally to the WBS common deletion
- Involved in neuronal homeostasis and signal transduction
- candidate genes for susceptibility to: Autism, epilepsy and intellectual disability

V) CONCLUSION:

- The importance to map precisely the WBS deletion.
- Evaluate genotype-phenotype correlation.
- Understand the gene function.

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