WILLIAMS SYNDROME - NEW CASE REPORT, EVALUATION AND MANAGEMENT

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LITERATURE DATA

Williams syndrome is a rare genetic disease caused by a 7q11.23 microdeletion.

Clinical phenotype includes:
- typical facial dysmorphism (*elfin* face),
- mental retardation,
- peculiar neuropsychological profile: relatively well-preserved language skills, visuospatial deficits and hypersociability,
- congenital heart defects.

Other features:
- neonatal hypercalcemia,
- ophthalmological anomalies,
- hypothyroidism,
- growth retardation,
- joint disturbances.

The inheritance is autosomal dominant but most cases are sporadic.

We present a case of a 10 years old patient with Williams syndrome in order to illustrate a rare disorder and to discuss the the management of the patient and her family.

HISTORY

- the girl is the first child born by an young, unrelated apparently healthy couple;
- pregnancy – uneventful;
- birth - natural, full term, low birth weight - 2500g;
- postnatal development - delayed

PHYSICAL EXAMINATION (10 years old)

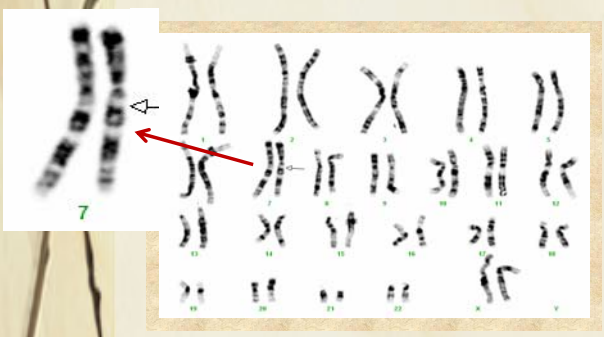
- hight stature (+2 SD), overweight (+3.28 SD)
- dysmorphic face:
  - elfin-like face,
  - stellate iris pattern,
  - flat nasal bridge with bulbous, upturned nose,
  - full lips,
  - wide mouth,
  - long philtrum;
- cubitus valgus,
- borderline intelligence.

INVESTIGATIONS

- echocardiography – bicuspid aortic valve, aortic, tricuspid and pulmonary insufficiency;
- renal ultrasound – normal;
- psychological examination - IQ 74.

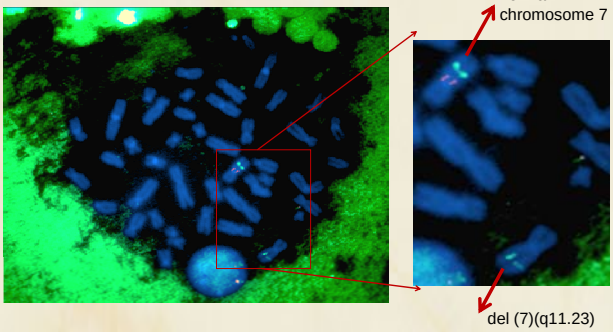



G TG band karyotyping: 46,XX,del(7)(q11.23;q11.23)



FISH analysis : 46,XX,ish del(7)(q11.23;q11.23)(ELN-).

- we use 7q11.23 LSI ELN - Spectrum Orange and 7q31 D7S486, D7S522 - Spectrum Green.



GENETIC COUNSELLING

- the karyotypes of the parents have been normal;
- recurrence risk is 1% (*de novo* deletion), but the possibility of gonadal mosaicism in one of the parents cannot be excluded;
- prenatal diagnosis is available.

MANAGEMENT

- no specific treatment is available,
- rehabilitation programs and educational interventions improve the prognosis and the social adjustment of the patients,
- yearly: medical evaluation, vision screening, measurement of blood pressure, calculation of calcium/creatine ratio in a random spot urine, and urinalysis,
- periodic evaluations: calcemia, thyroid function, hearing, renal and bladder ultrasound exam,
- diet (owerwwight, prevent hypercalcemia),
- during adulthood: glucose tolerance cardiac evaluation, ophthalmologic evaluation,
- surgery may be required for cardiac malformation.

CONCLUSIONS

- We present a a particular case of Williams Syndrome with borderline intelligence, complex heart disease, owerwieght and delayed diagnosis;
- Chromosomal analysis and FISH confirmed the deletion on chromosome 7;
- In our case we not found a correlation between the length of deletion and the phenotype, and this reflects the phenotypic variability in Williams syndrome
- Both child's and parental karyotypes are important for a correct diagnosis and genetic counselling