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.

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

INVESTIGATIONS

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

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 a particular case of Williams Syndrome with borderline intelligence, complex heart disease, overwieght 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

CONCLUSIONS

- 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.