Growth hormone deficiency in patient with ring chromosome 18

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The authors have nothing to disclose

Background
Ring chromosome 18 [r(18)] syndrome belongs to a rare group of chromosomal abnormalities. The clinical features of r(18) usually correlate with the sizes and locations of the delated genomic regions and the junction sites. Associated symptoms and findings may vary greatly in range and severity.
Patients with r(18) syndrome are characterized by short stature, obesity, microcephaly, mental retardation, hypertelorism, ptosis, epicanthic folds, micrognatia and small hands with short tapering fingers. Also atypical manifestations have been observed, with facial anomalies only.
The association of r(18) and growth hormone deficiency (GHD) is extremely rare (with only 3 descriptions in the literature with different responds to rGH treatment).

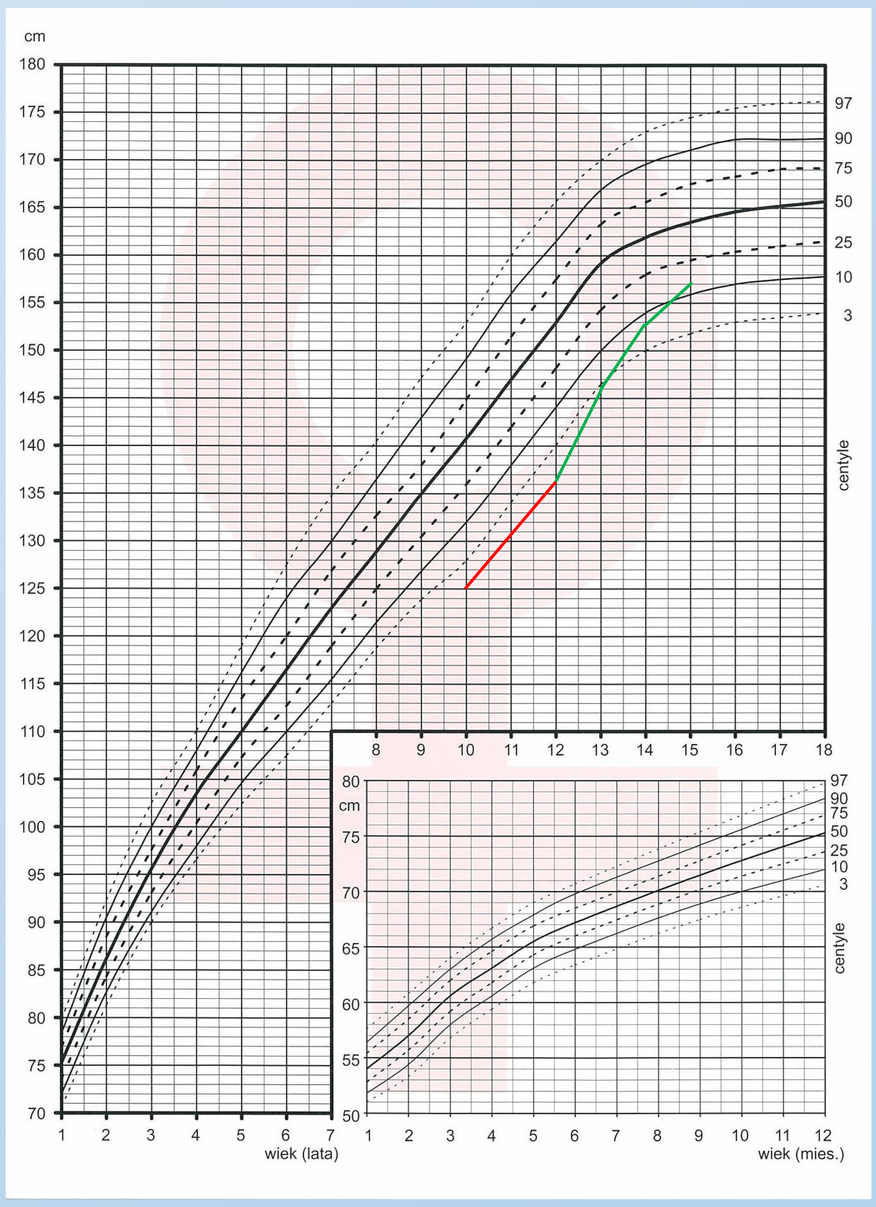
Case study:
A 12 years old girl
was referred to endocrinology department because of short stature.
She was born to healthy unrelated parents at 36 weeks of gestation.
Birth weight was 2200 g, length 51 cm, head circumference 32 cm, Apgar score 10.
There was no family history of genetic or congenital disorders. Her developmental milestones were normal.
She did not show any chronic diseases.

On presentation
Height 136 cm (-2,7 SDS), weight 40,6 kg, BMI 22.5 (1,1SDS)
Tanner stage: 2 breast, 1 pubic hair development,
discreet epicanthic folds, strabismus, lower-set ears and slightly smaller jaw. Small hands and feet.
Mental development was within normal range.

Laboratory
The diagnosis of GDH was made on the basis of GH stimulation tests.
GH peak response to clonidine: 2.69 ng/mL,
GH peak response to insulin: 3.61 ng/ml,
maximum spontaneous spurt of GH sampling during the night:
0,48 ng/ml.
IGF-1: 185 µg/ml, IGFBP3: 4,85 ug/l.
Baseline LH: 0,5 mU/l, FSH: 2,6 mU/l, TSH: 2,4 mU/ml, fT4: 14,2 pmol/l, prolactin: 198 mU/l, ACTH: 22 pg/l and cortisol: 287 pg/ml concentrations were within normal ranges for age.
TPO-Ab, TG-Ab, TR-Ab- negative ,Anti-GAD and ICA-negative,
LKM Ab-negative, AT-Ab (subclass IgA and IgG)- negative
Immunoglobulins (IgG, IgM, IgA)- normal rangers for age
Bone age - 10 years
Ultrasound of abdomen and thyroid – normal for age.
Echocardiogram showed minimal mitral regurgitation- clinically not relevant.

Magnetic resonance imaging of hypothalamic-pituitary region revealed a small (2mm) hypophysis.
Chromosome analysis showed:
46, XX.ish r (18)(wcp18x1)[11]

Treatment
rGH replacement therapy was introduced with the dose of 0,33mg/kg/day.
After the first year of treatment the girl reached height of 146,8 cm (-1,8 SDS) with the height increase Δ 0,9 SDS.
After the second year her height was 152,8 cm (-1,46 SDS), after the third: 156,5 cm (-1,1SDS).
Menarche occurred at the age 13 years 6 months.



CONCLUSIONS

Our patient's growth response to rGH treatment was similar to that observed in children with isolated growth hormone deficiency without r(18)syndrome.
In children with r(18) growth hormone deficiency should be excluded.

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