THE CHILDREN'S MEMORIAL HEALTH INSTITUTE
DEPARTMENT OF ENDOCRINOLOGY AND DIABETOLOGY

A patient with a 13q deletion syndrome, important growth delay and somatotropine insufficiency undergoing growth hormone therapy– case report.

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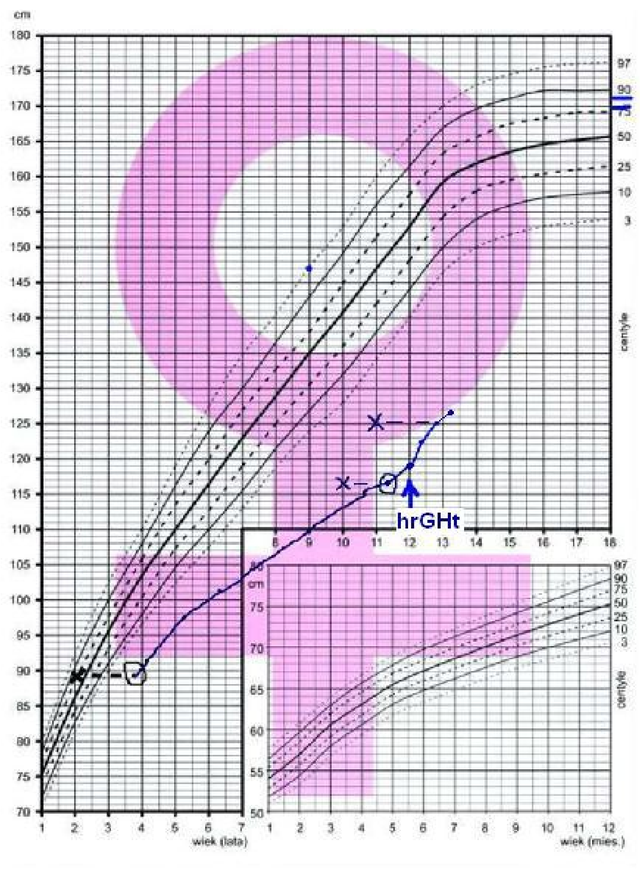
Growth deficiency is a common symptom of many genetic syndromes. 13q deletion is a very rare genetic syndrome described in less than 200 cases. Growth reduction is a constant symptom along with mental retardation and congenital defects varying according to the deleted region of chromosome 13. The aim of this paper was to present the case of a 12 year old patient with an interstitial deletion 13q (22.3-31.1), important growth delay and somatotropine insufficiency treated with hrGH.

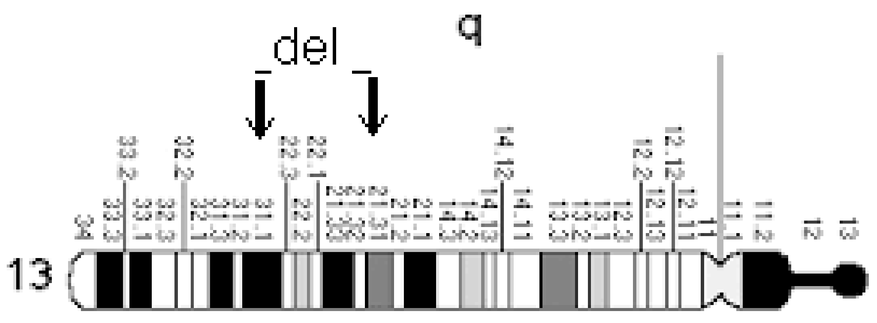
The patient is a first child of young non-consanguineous Polish parents. They had a miscarriage in the first trimester. The patient was born at term with 2550g weight (<10th c), 47cm length (10thc). In the neonatal period she showed failure to thrive, delayed psychomotor development and reduced muscle tone. Before one year of age she was diagnosed with 13q deletion syndrome (karyotype: 46XX, del 13q (22.3-32.?). She is a patient of numerous medical clinics for her diverse needs (dysgammaglobulinemia, fructose intolerance, Asperger Syndrome).

She has been a patient of the Endocrinology and Diabetology Department of the Children's Health Memorial Institute since 2005 (age: 3y). Growth delay is a constant symptom of the 13q deletion syndrome therefore hormonal tests haven't been performed at the time. Visits were discontinued from 2008 untill 2013, when she appeared presenting facial dismorphic features, scoliosis, lumbar hyperlordosis, weak postural and limb muscles, fifth toe clinodactyly, Ax1, Pub1, Th1, Me (-). **Height: 119,0cm, hSDS:-5.11, hSDS-mpSDS:-4.25, GV:2,7 cm/y, IGF-1:142ng/dl (N), BA: 9-10y.** GH secretion was low in 3 performed tests , i.e.: maximum GH concentrations were: in the night profile 3,63ng/ml, arginine test 0,9ng/ml and glucagone test 7,47ng/ml. Pituary MRI: dimensions AP 6mm, h: 3mm, transverse 12mm, without pathological enhance after gadoline administration. She was diagnosed with somatotropine deficiency. Other growth delay causes were excluded.



date	tests	results	units	Normal range
Tests evaluating growth hormone secretion				
26/27. 11.2013	Nocturne secretion	**0'**:0,24; **30'**:0,19; **60'**:1,28; **90'**:3,63; **120'**:1,8	ng/l	>10
26.11. 2013	L-Arguinine test	**0'**:0,8; **30'**:0,9; **60'**:0,89; **90'**:0,34; **120'**:0,26	ng/l	>10
27.11. 2013	Glucagone test	**0'**:0,18; **90'**:0,06; **120'**:1,2; **150'**:7,47; **180'**:2,39	ng/l	>10
30.08. 2013	IGF-1	142	ng/dl	188-515
18.03. 2015	IGF-1	96,9	ng/dl	214-753
27.08. 2015	IGF-1	216	ng/dl	214-753

With the permission of the Growth Hormone Treatment Coordination Group, in terms of a trial the patient was started with hrGH dosed 0,4U/kg/week (May-Nov.2014), and 0,34U/kg/week (Dec.-March2015), 0,35U/kg/week (March-May), 0,4U/kg/week (at present). After 10 months we achieved a GV improvment from 2,7 cm/y to 7,2cm/y and centile position change from **hSDS:-5.11 to hSDS:-4.89, ΔhSDS:0,22.** After 3 months of rhGH therapy the girl was supplied with orthopedic support preventing major back worsening.



In the Genetics Department of The Children's Health Memorial Institute a CGHarray assay showed **13q21.31-31.1 deletion** containing 34 genes, including Waardenburg type 4A Syndrome and Hirschprung Syndrome, of which she doesn't display the symptoms.

Conclusion: hrGH treatment might bring satisfactory results in children despite growth deficiency being a standard symptom in 13q deletion syndrome. This is the second reported patient undergoing rhGH treatment and responding to the treatment.

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