Skeletal Disproportion in Girls with Turner Syndrome

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Background and Aims

Untreated, Turner Syndrome (TS) results in a mean final height deficit of 21cm [1]. rhGH therapy can increase final height by 5cm, if commenced early [2]. Delayed diagnosis of TS still occurs [3,4].

In *SHOX* haploinsufficiency, children have disproportionately short limbs in relation to their spines [5].

The aims of this study are to

1) Evaluate sitting height and leg length in prepubertal girls with TS before rhGH
2) Evaluate sitting height and leg length with growth promoting therapies.

Methods

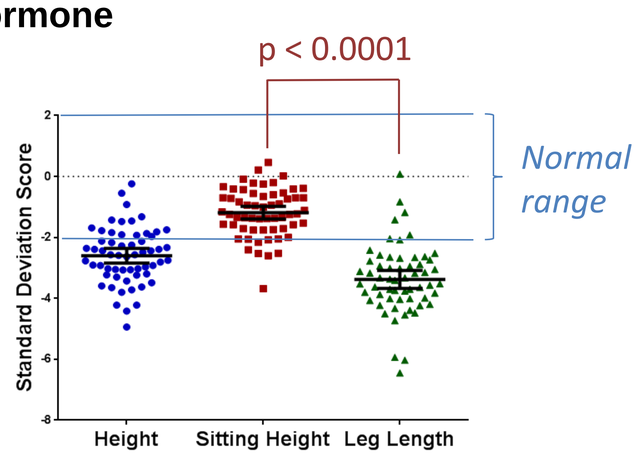
Retrospective study of clinic data from the West of Scotland TS clinic

Those with chronic disease potentially affecting growth were excluded eg. IBD, coeliac disease, thyroid disease, malignancy.

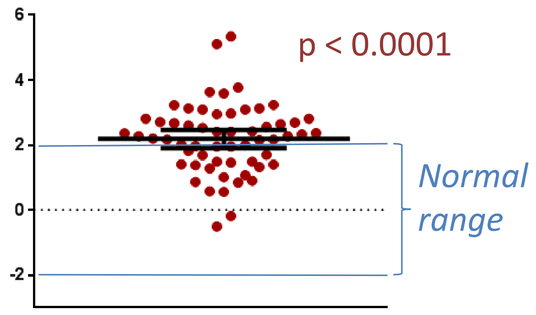
Results

Pre Growth Hormone

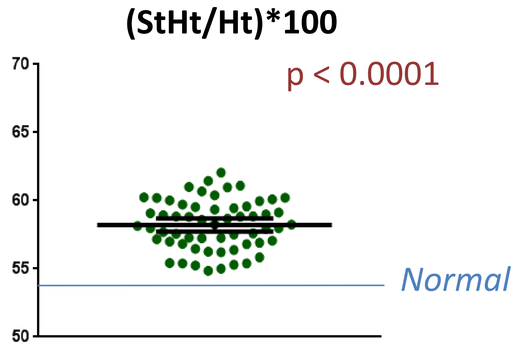
a)



b) StHt SDS – LL SDS



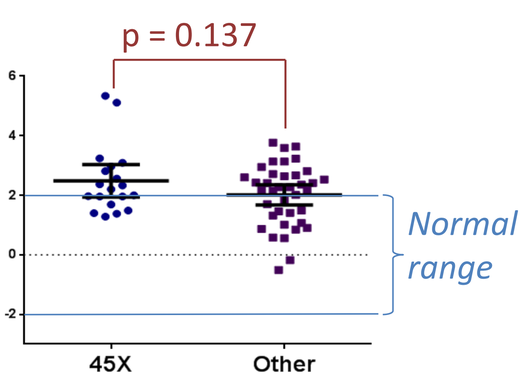
c) (StHt/Ht)*100



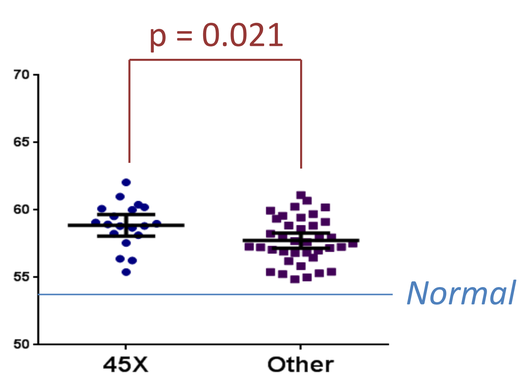
Figures a) to c) describe girls in the pre-growth hormone group (n=59). Median age = 6.1 years (range: 4.0 to 10.0 years). **a):** Height SDS, sitting height SDS and leg length SDS are all lower than average. Leg length SDS is significantly lower than sitting height SDS. **b) and c):** illustrate disproportion scores. Spinal length is significantly and disproportionately longer than leg length in each case.

Karyotype

d) StHt SDS – LL SDS



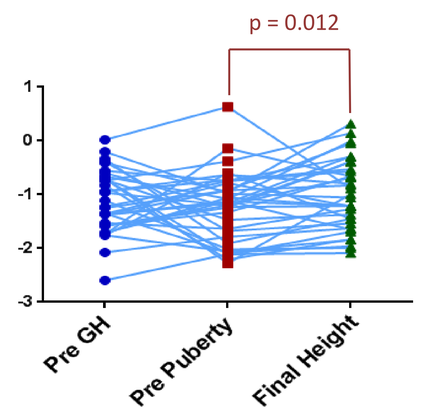
e) (StHt/Ht)*100



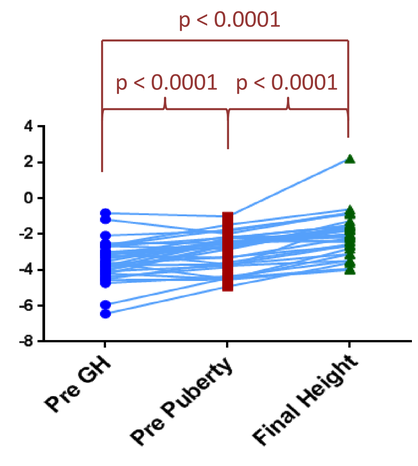
Figures d) and e) illustrate the effect of karyotype on disproportion scores, in pre-growth hormone children. 45X (n=19) is compared with all other karyotypes (n=38) . There is no significant difference between groups for sitting height SDS minus leg length SDS, although (sitting height (cm) / height)*100 is significantly larger in the 45X group, suggesting these children are more disproportionate.

Effect of Growth Hormone and Pubertal Induction

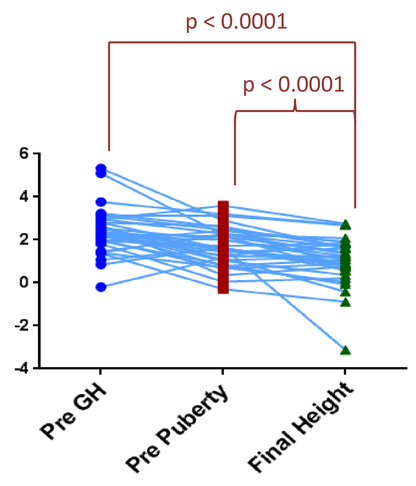
f) Sitting Height SDS



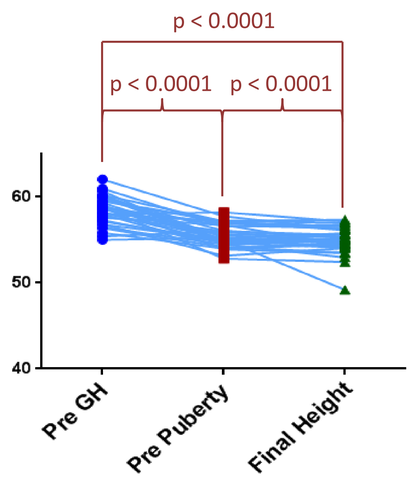
g) Leg Length SDS



h) StHt SDS – LL SDS



i) (StHt/Ht)*100



Figures f) to i) illustrate the change in standard deviation scores and disproportion scores from the pre-growth hormone phase in childhood to just prior to pubertal induction to final height.

Sitting height SDS does not change with childhood growth hormone therapy, but increases slightly at puberty.

Leg length SDS increases throughout both the childhood and the pubertal growth phases, and the children become significantly less disproportionate with treatment.

Summary and Discussion

(1) Prepubertal girls with TS have disproportionately shorter legs compared to their spine.

The diagnosis of TS may be more likely in a short girl with significantly shorter leg length.

(2) Girls with 45X may be more disproportionate compared with girls with other karyotype.

The effect of SHOX deficiency on skeletal disproportion in TS needs further investigation.

(3) Treatment with growth hormone and oestrogen reduce the skeletal disproportion in TS at final height.

Treatment factors and how they affect skeletal disproportion at final height should be studied.

References

1)Rochiccioli P, David M, Malpeuch G et al. Study of final height in Turner's syndrome: ethnic and genetic influences. *Acta Paediatr* 1994;83:305-8.

2)Betts PR, Butler GE, Donaldson MC et al. A decade of growth hormone treatment in girls with Turner syndrome in the UK. *Arch Dis Child* 1999;80:221-5.

3)Massa G, Verlinde F, Schepper JD et al. Trends in age at diagnosis of Turner syndrome. *Arch Dis Child* 2005;90:267-8.

4)Savendahl L, Davenport ML. Delayed diagnoses of Turner's syndrome: proposed guidelines for change. *J Pediatr.* 2000;137(4):455-9.

5)Binder G, Ranke MB, Martin DD. Auxology is a valuable instrument for the clinical diagnosis of *SHOX* haploinsufficiency in school-age children with unexplained short stature. *J Clin Endocrinol Metab.* 2003;88(10):4891-6.