Growth Hormone treatment in a child with Trisomy 21 and Turner Mosaicism

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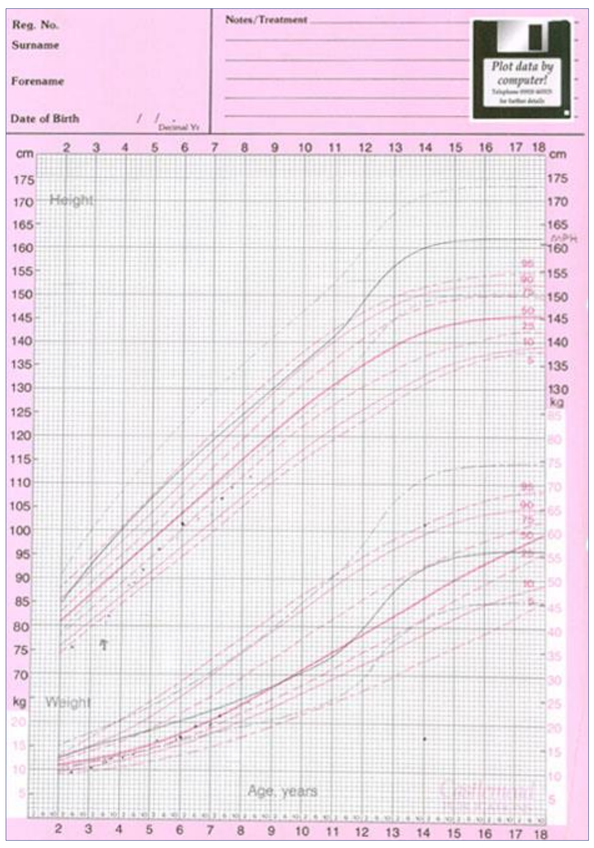
Background: Short stature occurs in Trisomy 21 but it is relatively slight during childhood. Turner syndrome would contribute significantly to short stature but the combined occurrence of both syndromes, even Turner mosaicism is unusual and could result in significant short stature.

Case: SP was referred for a growth assessment at 2.5 years. Her parents were counselled about short stature occurring in both syndromes but were worried that her height was significantly short even for either syndromes. Her height was 75.5cm (HtSDS -3.6 non Turner, HtSDS -1.8 Turner, <5th centile Downs), weight 9.26 kg (10th centile Downs). Facial features were consistent with Downs syndrome. The only Turner features were hyper convex nails. Her height velocity was noted to fall to 3.5 cm/year by 2.8 years, with height now at 76.8cm (HtSDS -4 non Turner, HtSDS -2.1 Turner), further away from the lowest centile of the Downs syndrome growth chart. The decision was made for a trial of growth hormone treatment and Turner doses with close monitoring of her IGF1 levels. She has now been on treatment for over 5 years. Auxology, IGF1 levels, GH dose are shown in the **Table**.

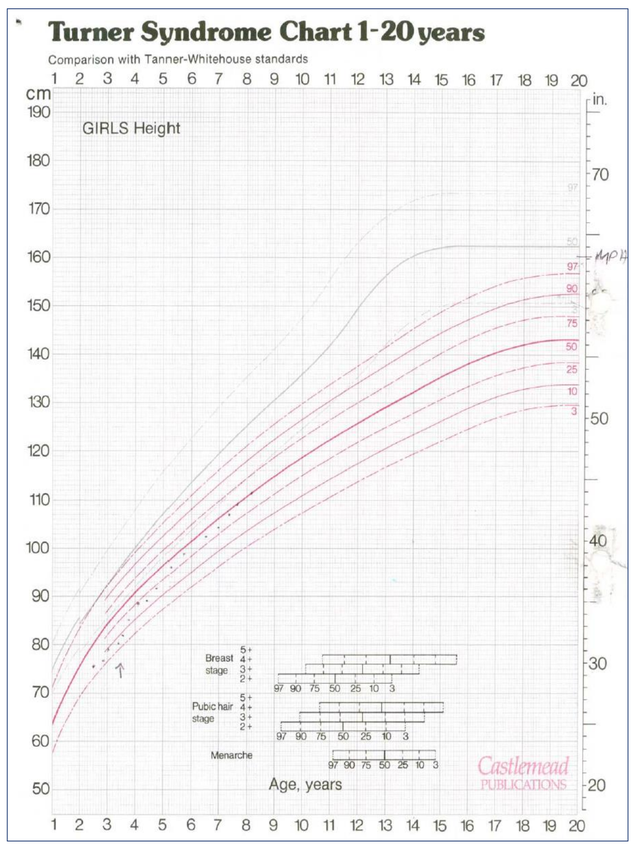
Investigations: Pre-treatment IGF1 171 ng/ml (51-303), TSH 1.79mU/L(0.4-4) fT4 9.8 pmol/L (6.3-14). Coeliac screen, Thyroid peroxidase antibodies negative. Renal ultrasound is normal. No cardiac lesions on echocardiogram. IGf1 11 months into treatment was 327 ng/ml (49-289), her GH dose was reduced when levels increased further.

Table

	7.4.10	3.12.10	21.3.11	31.10.11	11.11.13	13.8.14	16/2/15	6/11/15	6/5/16
HtSDS(Turner)	-2.01	-2.08	-1.95	-1.29	-0.57	-0.54	-0.64	-0.39	-0.32
HtSDS (nonTurner)	-3.97	-3.95	-3.84	-3.21	-2.59	-2.6	-2.7	-2.58	-2.57
IGF1					49.7 (3.3-31.7)	46.2 (3.3-31.7)	48.2 (5.1-48.2)	50.2 (5.1-48.2)	
GH (mg/m^2/wk)		4.2	7	8.4	9.5	5.8	4.9	5.6	6.4



Trisomy 21 growth chart



Turner Syndrome growth chart

Conclusion: Growth data over 5 years is presented in this unusual case on both Trisomy 21 and Turner growth chart. Both dysmorhpic syndromes are associated with short stature. Growth response to treatment has been good but dose titration critical due to concerns with raised IGF1 levels and Trisomy 21. Dose titration during pubertal induction would be most challenging, but it is reassuring to learn about the late effects of growth hormone treatment in Trisomy 21. (referenced).

References

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The author has nothing to disclose

55ESPE 867--P2

Syndromes : Mechanisms and Management

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DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS 10-13 SEPTEMBER

PosterSessionOnline.com