SENSITIVITY OF MEASURED PARENTAL HEIGHT AND TARGET RANGE IN THE DIAGNOSIS OF TURNER SYNDROME

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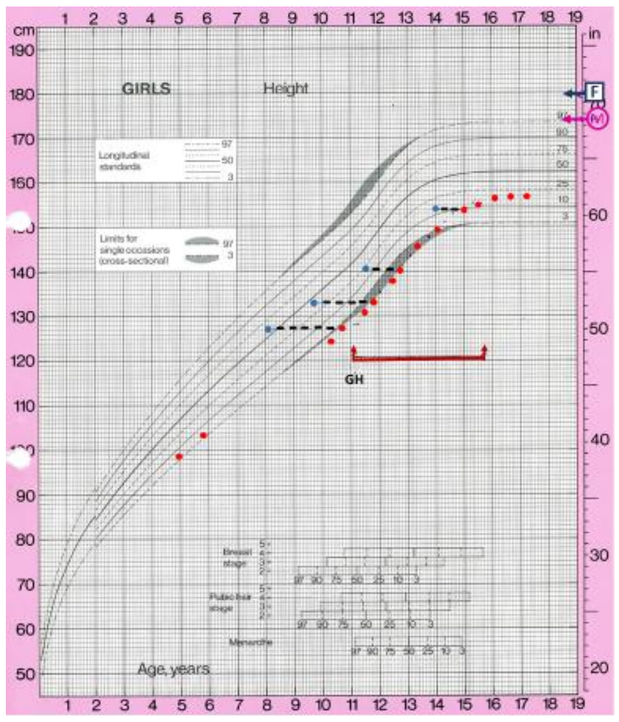
BACKGROUND

Girls with Turner syndrome are inappropriately short for their parents' heights (see growth chart opposite). Measured parental height is thus useful in diagnosis – but how sensitive is it?

AIMS

To examine the sensitivity of measured parental height in the diagnosis of Turner syndrome; and to audit the frequency of parental height measurement in our clinic.



METHODS

Case note review of all girls with Turner syndrome who had attended our dedicated Turner clinic between 1989-2013, recording the first accurate height measurement after the 1st year of life as well as karyotype, birth weight, gestational age and associated disorders. Each parent's height (Ht) was noted as measured, reported or not known/ not recorded. Ht and BW were converted to SDS using LMS software. Midparental height (MPH) and lower end of parental target range (LTR) were calculated using a 12.5 cm correction factor for gender, and 8.5 cm constant for 2 SD either side of MPH. Sensitivity of parental height measurement was calculated as the % of patients in whom Ht SDS (untreated and aged >1 year) was below LTR SDS.

RESULTS

Table 1. Demographic and descriptive information on 172 patients with TS seen at the Royal Hospital for Sick Children in Glasgow (1989 to 2013)

		All girls (n 172)	Both parents measured (n 94)	Only one parent measured (n 37)	Neither parent measured (n 41)
Age on 1.1.15	Mean (SD) Median (range)	26.28 ± 9.97 26.93 (3.34 – 54.24)	23.82 ± 8.80* 24.70 (3.73 – 38.83)	24.77 ± 8.11* 25.82 (8.99-38.61)	33.02 ± 10.54* 34.10 (3.34-54.24)
Age at initial measurement	Mean (SD) Median (range)	7.69 ± 4.64 7.69 (1.31 - 29.68)	6.83 ± 3.84 6.96 (1.31 - 15.49)	10.26 ± 5.85 11.39 (2.67 - 15.60)	8.53 ± 6.55 6.65 (1.39 - 29.68)
Height SDS at initial measurement	Mean (SD) Median (range)	-2.70 ± 1.07 (n 146) -2.69 (-6.30† to +1.79)	-2.63 ± 0.94 (n 92*) - 2.62 (- 4.71 to -0.32)	-2.67 ± 1.26 (n 36) -2.71 (1.79 / -6.30†)	- 2.92 ± 0.90 (n 19) -2.95 (-1.30 / -4.88)
Birthweight (grams) Median (range) [n]		2805 (690 – 4060) [n 136]	2800 (690 – 4060) [n 78]	3010 (1540 – 3860) [n 31]	2800 (1660 – 3660) [n 27]
Karyotype (n)		(n 170)	(n 92)	(n 37)	(n 39)
45,X		69	33	16	19
45, X/46XiXq		29	17	5	7
45,X/46,XY		9	6	3	0
45, X/46XX		9	4	3	1
45, X/47,XXX		11	7	4	0
45, X/ 46, X, r (X)		14	7	3	4
46, XiXq		6	5	1	0
Other		23	13	2	8

Table 2. Height standard deviation score (Ht SDS) of 92 girls with Turner syndrome in whom both parental heights were measured.

Pt Ht SDS	Group 1 1-5 yrs (n=38)	Group2 5.1-10 yrs (n=31)	Group 3 10.1-16 yrs (n=23)	Total 1-16 yrs (n=92)
<LTR SDS (n)	27	28	23	78
(%)	71	90.3	100	85
≥LTR SDS (n)	11	3	0	14
(%)	29	9.7	0	15

Table 3. Karyotype, height status and birthweight (BW) data in 14 girls with Turner syndrome whose initial accurate height measurement >1 year of age fell above the lower end of the parental target range (LTR) height SDS. Data are ranked according to age at initial height measurement

Karyotype	Age (yrs)	Ht SDS	LTR SDS	BW SDS	Comment
45,X	1.78	-1.52	-2.45	-0.02	Father's Ht <-2 SD
45,X	2.40	-1.42	-2.71	N/A	
45,X	2.46	-0.62	-1.25	0.09	
45,X/47,XXX	2.51	-0.60	-1.88	N/A	
45,X	2.69	-0.57	-1.43	N/A	
45,X/47,XXX	3.01	-0.75	-0.83	-2.74	
45,X	3.23	-1.25	-1.31	1.08	
45,X	3.27	-2.15	-2.78	1.98	Father's Ht <-2 SD
45,X/47,XXX	4.24	-1.19	-1.73	-1.76	
45,X	4.93	-2.15	-2.66	-1.30	
45,X/46,XX	4.99	-0.32	-1.72	N/A	
45,X/46,Xr(X)	7.20	-1.05	-1.46	N/A	
45,X/46,XX	9.08	-2.75	-2.85	N/A	
45,X	9.10	-2.02	-2.16	-1.34	

FINDINGS

- Measured height in both parents was available in only 94/172 (55%) girls
- Girls in whom neither parent was measured are older, suggesting that clinical practice has improved.
- Sensitivity of parental height measurement was 71% in girls aged <5 yrs, 90% in girls aged 5-10 yrs and 100% sensitive > 10 yrs
- The 14 girls whose Ht SDS was >LTR SDS were younger (11 were < 5 years), with milder genotype (45,X/46,XX and 45,X/47,XXX) in 5 girls.
- Of 136 girls with BW available 65(48%) had BW <-1 SD.

CONCLUSIONS

Evaluation of Ht status in the context of measured parental height is highly sensitive in girls with Turner syndrome aged >5 years and more specific than crude short stature screening. BW, although below average in >75%, is not a sensitive marker. Height status may be normal in younger girls and also those with milder karyotypes. This study reinforces the place of measured parental height in the growth clinic. Our experience indicates that this is still not happening consistently in paediatric practice.

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