PREVALENCE OF SCOLIOSIS IN A LARGE COHORT OF PEDIATRIC AND ADOLESCENT PATIENTS WITH PRADER-WILLI SYNDROME: A SCOTTISH-ITALIAN STUDY

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BACKGROUND

A variable prevalence of scoliosis has been reported in Prader-Willi syndrome (PWS) (40-90%). About 15% of patients with scoliosis develop severe curves, requiring bracing or surgery. Clinical detection can be challenging because these patients are obese and have less vertebral rotation than seen in other patients with scoliosis curves of a similar size, leading to a diagnostic delay. Marked generalized muscle hypotonia might influence the development or progression of scoliosis. The role of growth hormone (GH) therapy in the onset and progression of scoliosis remains controversial as does the modality of screening.

OBJECTIVES

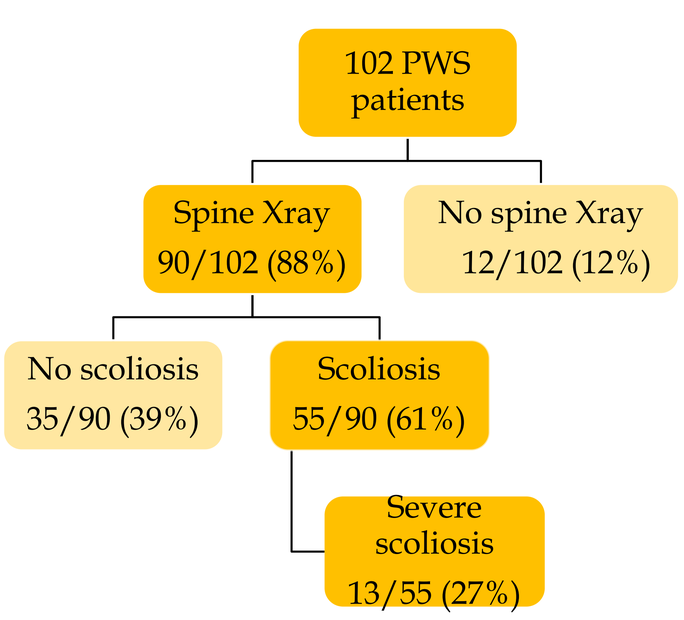
- to define the prevalence of scoliosis in our PWS patients
- to analyse the role of age, gender, genotype, BMI and GHT on its onset and severity.

METHOD

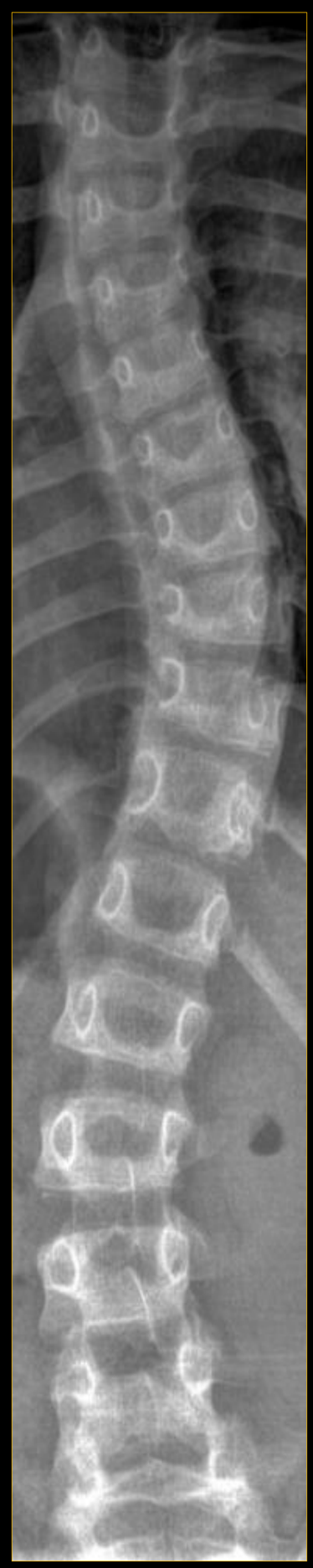
We analyzed patients 102 genetically confirmed PWS patients (44 deletion, 41 UPD, 17 unknown) attending Italian (n=74) and Scottish (n=28) PWS dedicated clinics between 2011 and 2014. Auxological and GH data was collected, as well as the assessment of scoliosis including Cobb Angles (CA) measurements. Scoliosis was defined with a CA>10°. Severe scoliosis (SS) was diagnosed with a CA>25° and requiring referral to spinal deformity service due to a high risk of progression.

RESULTS

- 102 PWS patients [50F] were studied, mean age 8.6 yrs (0.8-17.2). 89 (87%) patients are or have been on GH treatment. Spinal x-ray was performed in 90/102 (88%) patients. Scoliosis was present in 55/90 (61%) patients [31F, 24M] who had undergone x-ray. 13(27%) patients had SS.
- We did not find significant differences in age, gender, genotype or age starting GH among scoliosis and non-scoliosis group.
- In the scoliosis group, 81/89 (91%) patients were on GH treatment, previous or ongoing ($p<0.05$ vs non-scoliosis group; non-SS in 83%; $p<=0.001$ vs SS).
- BMI z-score was higher in scoliosis group (+1,3±1.3 vs -0.4±6.1, $p<0,05$).
- No difference was found among age, BMI z-score, age starting GH in SS vs non-SS patients.
- Univariate and multivariate analysis showed that only BMI z-score seems to influence scoliosis development (Beta:0.474; $p<0.001$), while age, gender and GH therapy did not seem to play a role. No correlation was found for SS patients.



	No scoliosis group (*n*=13)	Scoliosis group (*n*=89)
Mean age at follow up ±SD, y	12 ± 3.8	8.8 ± 4.8
Male gender, *n* (%)	6/52 (11,5%)	46/52 (88,5%)
Genotype, *n* (%) Deletion Disomy	 2/6 4/6	 42/79 37/79
Mean BMI z-score ±SD	-0.35 ± 6.16	+1.34 ± 1.31
GH treatment, *n* (%)	8/13 (61,5%)	81/89 (91%)
Mean age of starting GH ±SD, y	2,53 ± 1.81	3.00 ±2.83



CONCLUSION

In our patients scoliosis was frequent (61%), increasing with BMI z-score. It is difficult to define the role of GH, given the high percentage of our patients treated. We suggest regular spinal x-rays, especially where clinical spinal examination is difficult due to underlying obesity. Regular radiological assessment for scoliosis is justified pre and post-GH therapy.

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Miscellaneous
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