High prevalence of SGA in patients with disorders of sexual development (DSD), especially idiopathic 46,XY DSD

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Background

Disorders of sex development (DSD) are a group of rare conditions characterized by variable discordance between chromosomal, gonadal and phenotypic sex. An association between smallness-for-gestational age (SGA) and DSD is already recognised, but few studies have investigated this in detail.

Aim of the study

- To evaluate the prevalence of SGA, among patients with DSD
- To establish a correlation with the different types and causes of DSD.

Methods

All patients referred with DSD to our endocrine clinic from December 2007 to December 2015 were analysed in order to determine:

- the type of DSD (46,XY; 46, XX; or sex chromosome DSD) and where possible the precise aetiology based on clinical assessment, and hormonal, radiological and genetic investigations.
- SGA status as defined by birthweight (BW) or length (BL) < 10th centile according to the Audipog database. Low birth weight (LBW) was defined as BW <2500g

Statistical analysis was carried out using Epi-info7 and BiostaTGV.

Results

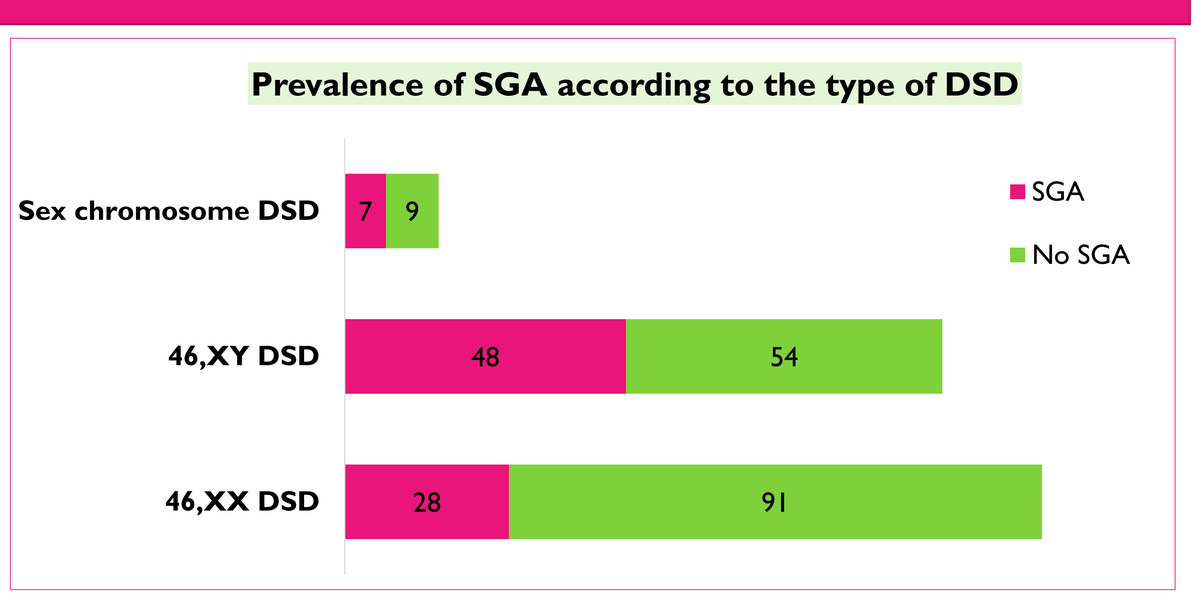
During the study period 237 patients were referred with DSD: 119 patients with 46,XX DSD, 102 with 46,XY DSD and 16 patients with sex chromosome DSD.

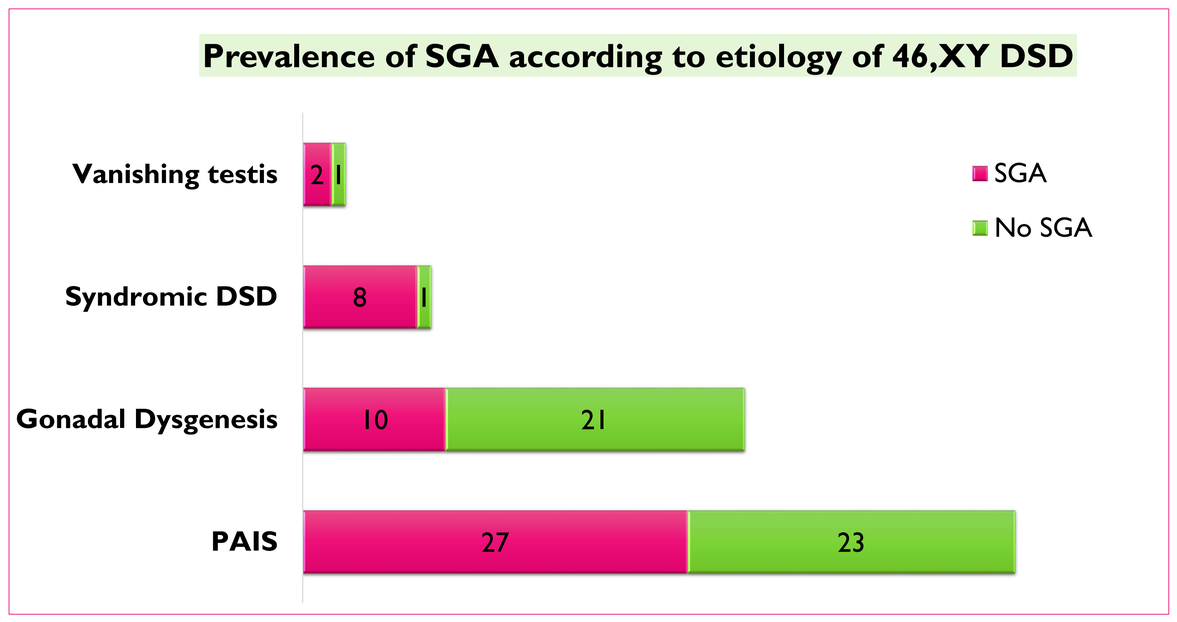
Median (range) birth weight was 3.1(2.66-5) kg, birth length 49.5 (38-57) cm and gestation 40 (25-42) weeks.

SGA was present in 83 (35%) infants, 45 (54%) of whom had BW <2500 g.

The prevalence of SGA was higher in the groups with 46,XY DSD (47%) [n=48] and with sex chromosome DSD (44%) [n=7], than in the 46,XX DSD group (23.5%) [n=28] ($p<0.01$).

In the 46,XY DSD group, the prevalence of SGA was particularly high in patients with syndromic DSD (89%) [n=8] and AR mutation-negative partial androgen insensitivity syndrome (54%) [n=27] ($p<0.05$). Within this latter group there was no difference in the prevalence of SGA according to the EMS score.





Conclusions

This study confirms the association between SGA and DSD[2], which appears especially strong in patients with idiopathic 46,XY DSD[2,3,4].

The question remains as to whether there is a common genetic mechanism causing both DSD and SGA in 46,XY patients; or if the defect in prenatal exposure to androgens *per se* affects intrauterine growth.

References

[1] *www.Audipog.net*

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Gonads & DSD

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