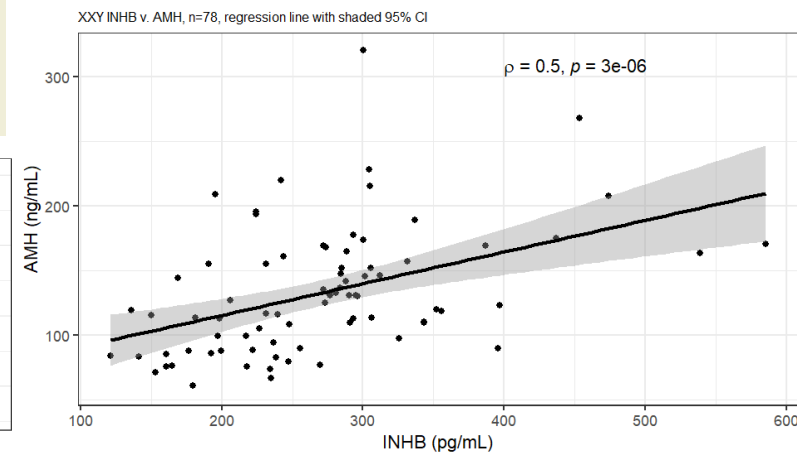
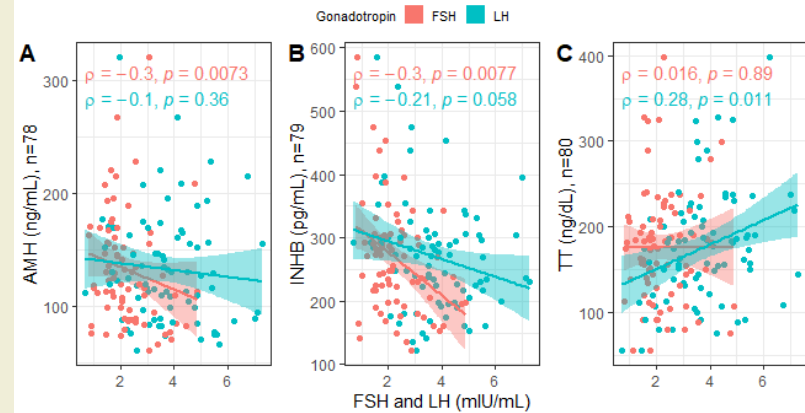
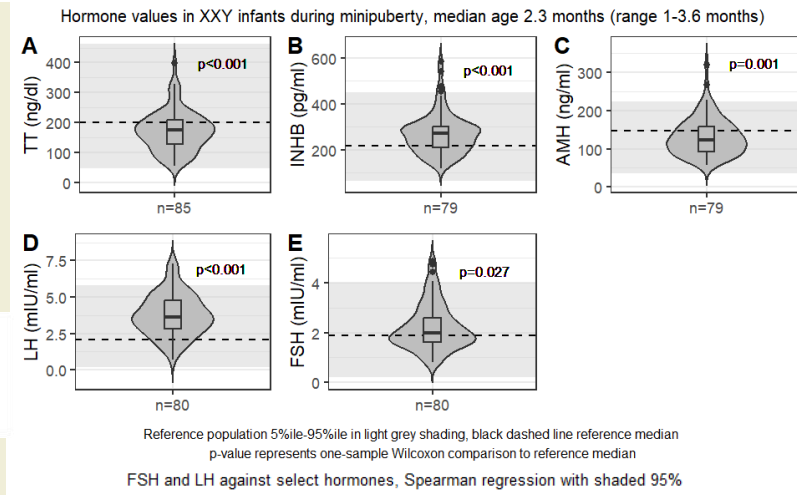


Minipuberty pituitary-gonadal hormone profile in infants with 47,XXY

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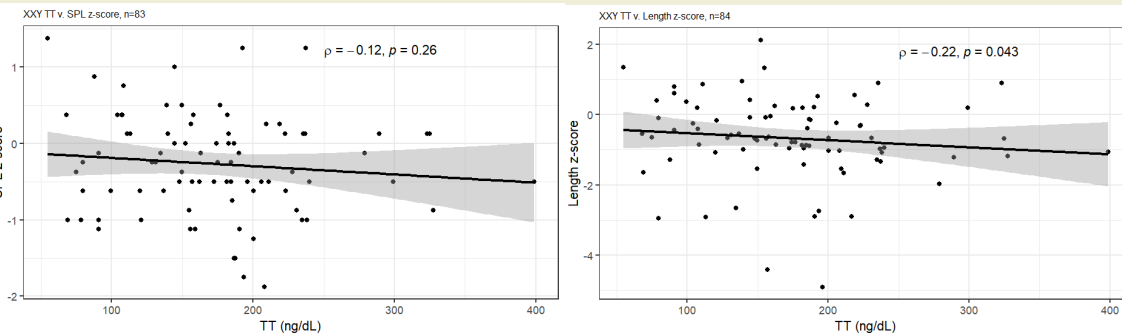


Background and Aims

- Activation of the hypothalamic-pituitary-gonadal axis in infancy, known as minipuberty, is important for growth of the phallus, maturation of the testes, early body composition, and potentially later metabolic and neurodevelopmental programming
- A systematic review of the previous studies of minipuberty in males with 47,XXY Klinefelter Syndrome (KS) included a total of only 87 infants and was limited by disparate hormone measurement techniques
- Our aim was to describe systemic gonadal hormone concentrations in infants with KS during minipuberty

Methods

- 85 infant XXY males participating in the eXtraordinary Babies and/or TESTO studies
- Serum hormone concentrations drawn during minipuberty (median age 2.3 months)
- Total testosterone (TT) was analyzed using ultra-high performance liquid chromatography tandem mass spectrometry. Luteinizing hormone (LH), follicle stimulating hormone (FSH), inhibin B, and anti-mullerian hormone (AMH) were measured via immunoassays
- One-sample Wilcoxon using the reference population medians were performed for violin plots, Spearman regression analyses performed for scatterplots with alpha set at 0.05



Results

- Most hormone concentrations fell within normal ranges, however the median levels were lower for TT and AMH and higher for INHB, LH, and FSH compared to the reference sample.
- Positive correlations observed between TT/LH and AMH/INHB
- Negative correlations observed between AMH/FSH and INHB/FSH
- No correlation between TT and stretched penile length z-score, weak negative correlation between TT and length z-score

Conclusions

- Group differences suggest differential hormone production in infants with XXY compared to normative values
- Lack of positive correlation between TT and physical measurements challenges physiologic implication of TT in this period

Implications & future directions

- Contradictory finding between FSH and INHB compared to previous studies in KS infants that showed a positive correlation
- Strengths of this study include uniform hormone measurement techniques, a large sample size, and a narrow age window compared to previous studies
- Limitations of this study include not controlling for factors like SES, maternal health, complications during pregnancy and/or labor, and prematurity.
- Can hormone measurements in minipuberty predict longitudinal health or developmental outcomes in this population?

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