

Ultrasound-guided biopsy of the submental area showed cytomorphic features of a colloid nodule with cystic degeneration (Bethesda Category II). The patient was started on levothyroxine and remained biochemically euthyroid afterwards. The submental neck mass reduced in size.

Conclusion: Dual ectopic thyroid with normally located (eutopic) thyroid gland could present with subclinical hypothyroidism. There is no single diagnostic modality that would best identify dual ectopic thyroid; thus, thyroid scan, ultrasonography, CT scan and biopsy are recommended to be used complementarily. For patients with dual ectopic thyroid and hypothyroidism, levothyroxine replacement is recommended to reduce the size of ectopic thyroid and render the patient euthyroid.

Pediatric Endocrinology

PEDIATRIC PUBERTY, TRANSGENDER HEALTH, AND GENERAL ENDOCRINE

Steroid Hormone Profile Differentiates Gynecomastia and Pseudo- Gynecomastia in Pubertal Boys

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SUN-060

Background: Gynecomastia (defined by breast tissue) and pseudogynecomastia (defined by adipose tissue) is frequent in pubertal boys. However, the underlying pathomechanisms are not fully understood so far. An association to growth hormone axis- IGF-1 axis and sex hormones has been discussed. **Methods:** We analyzed serum steroid hormones (progesterone, estradiol [E2], estriol, estrone, cortisol, cortisone, testosterone [T], dihydrotestosterone [DHT]) by liquid chromatography-tandem mass spectrometry, as well as gonadotropins, prolactin, IGF-1 and IGFBP-3 in 124 pubertal boys with breast swelling (mean age 14 +/-2 years). The steroid hormones were compared to those of 84 healthy pubertal boys (mean age 14 +/-2 years) without breast swelling. The differential diagnosis of either gynecomastia or pseudogynecomastia was determined by ultrasound. Puberty was defined by testes volumes > 3ml on each side. **Results:** A total of 86 boys suffered from gynecomastia and 38 from pseudogynecomastia. In boys with gynecomastia the ratio E2/T (median 22, interquartile range [IQR] 8–75) was significantly ($p<0.05$) higher compared to boys with pseudogynecomastia (median 12 IQR 5–21) or healthy boys without breast swelling (median 18 IQR 6–44). DHT concentrations were significantly ($p<0.001$) lower in boys with gynecomastia (median 0.13 IQR 0.02–0.38 nM/L) or pseudogynecomastia (median 0.18 IQR 0.05–0.32 nM/L) compared to healthy boys (median 0.41 IQR 0.22–0.66 nM/L). T concentrations were significantly ($p<0.05$) lower in boys with gynecomastia (median 1.8 IQR 0.7–4.2 nM/L) compared to boys with pseudogynecomastia (median 4.3 IQR 1.4–6.9 nM/L) or healthy boys without breast swelling (median 3.1 IQR 0.6–7.6 nM/L). The ratio DHT/T was significantly ($p<0.001$) lower in boys with gynecomastia (median 0.09 IQR 0.02–0.17) or pseudogynecomastia (median 0.04

IQR 0.02–0.16) compared to healthy Boys without breast swelling (median 0.13 IQR 0.05–0.28). Boys with gynecomastia did not differ from boys with pseudogynecomastia according to the other steroid hormones, prolactin, IGF-1, or IGFBP-3 concentrations. **Conclusions:** Gynecomastia is characterized by a higher E2 to T ratio compared to healthy boys without breast swelling due to a relative T deficiency in the presence of similar E2 levels. The lower DHT/T ratio in gynecomastia and pseudogynecomastia compared to healthy boys without breast swelling points towards a functional 5 alpha reductase deficiency.

Genetics and Development (including Gene Regulation)

GENETICS AND DEVELOPMENT AND NON-STEROID HORMONE SIGNALING II

Transcriptome Profiling in Postnatal Pituitary Gland Identifies Cell Type-Driven Sex-Specific Changes

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MON-725

The pituitary gland is integral to the regulation of growth, metabolism, puberty, reproduction, and stress responses. Previously, we found that many genes associated with age-at-menarche in genome-wide association studies (GWAS) displayed increasingly sex-biased expression across the pubertal transition in the mouse pituitary. However, whether this trend exists beyond puberty-related genes was not known. In addition, the regulatory mechanisms underlying these gene expression changes remained to be explored. To answer these questions, we profiled the transcriptome, including microRNAs, of mouse pituitary in both sexes across pubertal transition in an unbiased manner and leveraged a recently published pituitary single cell transcriptome to explore cellular composition changes. We found that the most dynamic temporal changes in both mRNA and miRNA expression occur prior to puberty, underscoring a role for regulation of early pituitary postnatal development. We also observed ~900 genes displaying sex-biased expression patterns, arising during early development and becoming increasingly biased across puberty, including known sex-biased genes such as *Fshb* and *Lhb*. However, sex differences in miRNA expression are less pronounced, only 13 miRNAs were found to be sex-biased, suggesting lesser contribution of miRNAs to sex-biased gene expression relative to other forms of regulation. To assess whether pituitary cellular composition could underlie changes in gene expression across pubertal transition, we performed single cell deconvolution of our bulk pituitary gland gene expression. Interestingly, we found that sex differences in cell proportions were estimated to emerge across puberty: a greater proportion of lactotropes was found among females, and greater proportions of gonadotropes and somatotropes were found among males. We observed sex-biased expression patterns of marker genes for these cell types, including *Prl*, *Fshb*, and *Gh*. This finding suggests that cell proportion