

Ordering pitMRI when PRL/T >0.08 or when PRL >25 is 98% sensitive (39/40 lesions captured) and 32% specific (32/101 with normal anatomy excluded). 108/141 are indicated for pitMRI, while 33 patients avoid imaging. Employing this threshold reduces expenses by 21% with cost savings calculated at \$96,195. The cost of identifying each lesion was estimated at \$9,011.

Conclusions: Serum PRL and PRL/T correctly predict the vast majority of pituitary lesions in patients with mild hyperprolactinemia, with screening costs increasing as more sensitive thresholds are employed. Future guidelines should establish a reasonable cutoff for pitMRI to minimize the expense of unnecessary imaging.

Steroid Hormones and Receptors

STEROID AND NUCLEAR RECEPTORS

Functional Analysis of Testis-Specific Noncoding Genes in Estrogen-Dependent Transcription

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Emerging studies have shown that germ cell (GC)-specific genes play critical roles in several cancers. The expression of these genes is tightly regulated and restricted to testis; however, many of them escape regulation and become aberrantly expressed in tumors. Interestingly, our genomic analysis suggests that several of these genes are long noncoding RNAs (lncRNAs) and are located at regions previously considered to be gene deserts in the human genome. In this regard, we used an integrated genomic approach to identify GC-lncRNA genes that are overexpressed in breast cancer. Further, by incorporating gene expression analysis from RNA-seq data from MCF-7 and T47D breast cancer cells, we generated a comprehensive list of estrogen-regulated GC-lncRNA genes. We hypothesize that GC-lncRNA genes regulate estrogen-dependent signaling in breast cancer. The selected genes: (a) CAERRC (Chromatin Associated Estrogen-Regulated RNA in Cancer), (b) LncRNA568, (c) LncRNA16 are primate-specific, and exclusively expressed in testis. All of them are regulated by estrogen, and their expression predicts poor outcome in ER α + breast cancer patients. They have now been fully annotated (transcription start and stop site, 5' cap, polyA tail, and exon/intron structure), and cloned. Further, we have created gene-specific KO MCF-7 cell lines using CRISPR to study their molecular roles. Our data suggest that these genes regulate estrogen-dependent gene expression and tumor growth in breast cancer cells. Genome-wide analysis of ER α binding and gene expression data indicate that they play a critical role in the estrogen-dependent transcription. Collectively, our results suggest that GC-genes, including CAERRC, LncRNA568, and LncRNA16, are excellent targets with prognostic and therapeutic potential in ER+ breast cancers.

Pediatric Endocrinology

PEDIATRIC SEXUAL DIFFERENTIATION, PUBERTY, AND BONE BIOLOGY

Pilot Study Using Aromatase Inhibitor in Puberty of Boys With Partial Androgen Insensitivity: Report of Three Cases.

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Background: Partial Androgen Insensitivity Syndrome (PAIS) (OMIM # 312300) is one of the causes of Disorders/Differences of Sex Development with 46,XY karyotype and normal or increased testosterone secretion that results in atypical genitalia. In general, it is caused by inactivating mutations on AR gene (Xq12 - OMIM * 313700), therefore it presents an X-linked recessive inheritance. Individuals raised as males have incomplete puberty with micropenis, sparse hairs, gynecomastia and increased LH, testosterone and estradiol serum levels. Therefore, the use of aromatase inhibitor in these cases becomes a logical indication aiming to increase testosterone levels in the tentative of supplanting its peripheral resistance and decreasing estrogen levels. **Objective:** To present clinical and laboratory data during the first year of use of aromatase inhibitor in puberty of three boys with confirmed molecular diagnosis of PAIS. **Results:** All subjects used letrozole (2.5 mg daily during 12 months). None reported significant side effects. Cases 1 and 2 are brothers (p.Ala596Tre). **Case 1:** Onset of treatment at age 12; height changed from 158 cm (z = +1.15) to 166 cm (z = +1.21), Tanner from G2P2T2 to G3P3T1, penis from 4.0 to 6.5 cm, LH from 7.5 to 18.3 IU/L (NR: 1.5 to 9.3 IU/L), testosterone from 361 to 1,347 ng/dL (NR: 165 to 763 ng/dL), estradiol from 35 to <5.0 pg/mL (NR: < 40 pg/mL) and bone age remained at 13 years. **Case 2:** Onset of treatment at age 11; height changed from 156 cm (z = +1.71) to 163 cm (z = +1.80), Tanner from G2P2T2 to G3P3T1, penis from 3.5 to 5.5 cm, LH from 4.8 to 12.4 IU/L (NR: 1.5 to 9.3 IU/L), testosterone from 259 to 1,069 ng/dL (NR: 165 to 763 ng/dL), estradiol from 28 to <5.0 pg/mL (NR: < 40 pg/mL) and bone age remained at 12.5 years. **Case 3** (p.Ser597Arg): Onset of treatment at age 12; height changed from 153 cm (z = +0.49) to 161 cm (z = +0.55), Tanner from G2P2T1 to G3P3T1, penis from 4.0 to 7.0 cm, LH from 8.5 to 17.2 IU/L (NR: 1.5 to 9.3 IU/L), testosterone from 280 to 889 ng/dL (NR: 165 to 763 ng/dL), estradiol from 32 to <5.0 pg/mL (NR: < 40 pg/mL) and bone age remained at 13.5 years. **Discussion:** There are no reports in the literature of the use of aromatase inhibitor in PAIS. The results of this pilot study (gynecomastia regression, significant testosterone level increases with decrease in estradiol levels, increment in height without bone age advancing, progression of puberty and penile growth) are sustaining for the indication of this treatment in boys with PAIS during puberty.