1) the adult endocrinologist should carefully read paediatricians’ letters and check whether action is required (i.e. check whether an appointment is requested)
2) the paediatrician should ascertain whether the appointment is really made and received by the patient
3) the patients and/or caregivers should be instructed to alarm the hospital when they do not receive the appointment.
These actions require relatively little effort and may prevent the part of drop-outs that is caused by logistic failures.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS I
Twins with a Homozygous Variant of ARNT2, This Is a Known Saudi Mutation (KSM) of Webb-Dattani Syndrome
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SAT-062
Webb-Dattani syndrome (WEDAS) is an autosomal recessive disorder caused by mutation in the ARNT2 gene characterized by frontotemporal hypoplasia, globally delayed development, and pituitary and hypothalamic insufficiency. The condition is reported to be associated with consanguinity and with Saudi Arabian ancestry. We presented twin baby girls with developmental delay seizures, and microcephaly. They have also hypopituitarism in the form of diabetes insipidus and hypocortisolism. also they have cortical blindness. Their brain MRI shows brain atrophic changes and delayed myelination thin corpus callosum, and small pituitary gland ad absence posterior high signal spot and pituitary stalk. Genetic testing by Exome sequencing was done and it shows A homozygous variant of ARNT2 (ARNT2:NM_014862:exon3:c.147-1G>A). One of this twin her condition deteriorated with uncontrolled seizures and spasticity and died at age 22 months. Conclusion: we report another cases of the ARNT2 mutation in a Saudi family illustrating the disease of webb-dattani Syndrome with seizures and hypopituitarism and severe visual impairment and global developmental delayment.

Tumor Biology

TUMOR BIOLOGY: GENERAL, TUMORGENESIS, PROGRESSION, AND METASTASIS

The Secretory Vesicle Membrane Protein, CYB561, Promotes the Growth and Metastatic Potential of Castration-Resistant Neuroendocrine Prostate Cancer
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SAT-132
An increase in the population of neuroendocrine (NE) differentiated (NED) cells and their secretory products are closely correlated with prostate cancer (PCa) resistance to existing therapies and eventual progression to castration-resistant PCa (CRPC). It is hypothesized that NED cells secrete neuropeptides that support tumor growth and induce aggressiveness of adjacent proliferating tumor cells through a paracrine mechanism. A gene that is constitutively expressed in secretory vesicles of NE cells, and has been previously found to be highly expressed in CRPC and cancer of several tissues is Cytochrome b561 (CYB561). The CYB561 gene encodes a secretory vesicle transmembrane protein that primarily functions in the regeneration of ascorbic acid, a necessary step in the α-amidation activation process in the biosynthesis of most neuropeptides. The CYB561 protein also exhibits ferrireductase activity and may contribute in regulating iron transport and metabolism, which are two other pathways often dysregulated in cancer. These findings led us to hypothesize that CYB561 may be a key player in the NE differentiation process that drives the progression of prostate cancers into the more aggressive NE subtype. In our study, we found that CYB561 expression is higher in metastatic and NE PCa (NEPC) models compared to normal prostate epithelia, and that its expression is not affected by androgen treatment or steroid deprivation. Lentiviral-mediated knockdown of CYB561 in the NEPC cell line, PC-3, decreased the expression of genes involved in NE differentiation and labile iron pool storage, decreased cell proliferation, reduced cell survival in a colony formation assay, and slowed down cell migration in a wound-healing assay. Treatment of normal prostate epithelial cells, PNT1A, with conditioned media from CYB561 knockdown PC-3 cells led to a decrease in proliferation rate when compared to treatment of PNT1A cells with media from CYB561 expressing (control) PC-3 cells. Taken together, our findings demonstrate the role of CYB561 in supporting the growth and metastatic potential of NEPC cells, and highlights the potential use of CYB561 as a therapeutic target and biomarker that can be used to identify more aggressive disease.