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  
Abdullah Abdulrahman Aljasser, MD.  
Prince Sultan Military Medical City, Riyadh, Saudi Arabia.

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 hypothyroidism, 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 delay.

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  
Kevin Christian V. Olarte, M.Sc., Pia D. Bagamasbad, PhD.  
National Institute of Molecular Biology and Biotechnology, University of the Philippines Diliman, Quezon City, Metro Manila, Philippines.

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 PCas (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 PCas (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.

Pediatric Endocrinology  
PEDIATRIC ENDOCRINE CASE REPORTS II  
A Case of Growth Hormone Deficiency in Sturge-Weber Syndrome  
Jeongju Hwang, MD, Jeesuk Yu, MD, PhD.  
Dankook University Hospital, Dankook University College of Medicine, Cheonan, Korea, Republic of.

MON-077  
Introduction: Sturge-Weber syndrome (SWS) is a congenital neurocutaneous disorder characterized by a port wine stain on the skin in the distribution of the ophthalmic branch of the trigeminal nerve (vascular malformation of skin), glaucoma, and leptomeningeal angiomas. Central nervous system abnormalities may increase the risk of hypothalamic-pituitary dysfunction. One previous study showed that SWS patients had higher prevalence of growth hormone deficiency than the general population although the etiology is unclear. This case report describes a patient who was initially diagnosed with SWS and later confirmed with complete growth hormone deficiency.  
Case: A 7-year-and-11-month-old boy who had been diagnosed with SWS visited a tertiary center for the
evaluation of short stature [111.1 cm (<3 percentile)]. He was born at 37 weeks of gestational age with birth weight of 3230 g by cesarean section and had a port wine nevus on the right side of the face since birth. He had a history of epilepsy first occurring at 6 months of age when he was diagnosed with SWS. There were two more attacks of seizure till 32 months of age. Brain magnetic resonance imaging revealed leptomeningeal angioma, choroidal hemangioma, and diffuse brain atrophy. He was diagnosed with glaucoma and had been managed with surgery and medication. There was no family history of SWS or any other brain anomaly. His mid-parental height was 167.7 cm. All blood tests were normal including complete blood count, chemistry, and thyroid function test. Hand x-ray showed delayed bone age. Cocktail test was performed for the evaluation of short stature. As a result, he was diagnosed with complete growth hormone deficiency (peak GH on L-dopa test: 2.46 ng/mL, peak GH on glucagon test: 3.71 ng/mL). The recombinant growth hormone therapy was started at the age of 8 years and 1 month. He showed good response to GH treatment. His height became 125.8 cm at the age of 9 years and 5 months (height velocity 9.2 cm/year), and 134.3 cm at the age of 10 years and 6 months.

Conclusion: We experienced a case with Sturge-Weber syndrome and complete growth hormone deficiency which was successfully managed by recombinant growth hormone therapy. It may be better to consider the possibility of GH deficiency even if there are certain conditions that affect the growth itself.

Neuroendocrinology and Pituitary
CASE REPORTS IN UNUSUAL PATHOLOGIES IN THE PITUITARY

Significant Response to Temozolomide in Two Aggressively Growing Pituitary Adenomas
Ilonka Kreitschmann-Andermahr, Prof. Dr. med., Agnieszka Gryzwoz, Dr. med., Claudia Möller-Hartmann, Dr. med., Andreas Junker, Dr. med., Dagmer Führer-Sakel, Prof. Dr. med., Nicole Unger, PD Dr. med.
University Hospital Essen, Essen, Germany.

SUN-272
Introduction: Aggressive atypical pituitary tumors are characterized by invasive growth, recurrence and resistance to standard therapies. We present two female patients with pituitary adenomas in whom multiple other therapies had failed, who presented with significant response to temozolomide. Case presentations: In patient #1 (w, 78y), the diagnosis of macroadenoma had been made in a community hospital and dopaminagonistic treatment with bromocriptin had been initiated. After failure to achieve significant tumor reduction under this treatment and persisting visual field disturbances, first transnasal-transphenoidal surgery (TSS) was performed in 07/2011, followed by cabergoline exposure in increasing dose due to failure to control prolactin levels. Repeat TSS and stereotactic radiosurgery were performed in both 2014 and 2018 because of invasive tumor growth and double vision. She was then put on temozolomide. Patient #2 (w, 58y) presented with apolectic gonadotropinoma in 2013. She also underwent 3 courses of TSS as well as stereotactic radiosurgery because of repeated tumor growth leading to visual field disturbances and double vision. Despite these measures, the tumor could not be controlled and she, as well, was put on temozolomide in 2018. In both cases costs were reimbursed by the patient’s health care insurance and in both the first cycle was conducted with 150 mg/ body surface area (BSA) with escalation to 200 mg/BSA in the second. After only 2 cycles, double vision resolved in both patients and the tumor had shrunk by approximately 20% on MRI in patient #1 and even more in patient #2. In both patients, temozolomide dose was reduced again to 150 mg/ BSA due to side effects. Nevertheless, in both patients tumor volume further continued to decrease under therapy. Conclusion: This promising clinical course after exposure to temozolomide with early, significant tumor shrinkage in two heavily pretreated patients with aggressive pituitary adenomas indicates that this therapy can be considered also in older patients and may yield astonishing results. Although temozolomide is increasingly becoming a therapeutic option for those patients whose pituitary tumors are refractory to standard therapies, further research and observation over time of temozolomide therapy in aggressive pituitary adenomas and carcinomas is indicated.

Pediatric Endocrinology
PEDIATRIC ENDOCRINE CASE REPORTS II

46 XX DSD Due to POR Deficiency
Nithin Modhugu Reddy, MD, Bipin Kumar Sethi, MD,DM.
Srinivas G.N.S.V. Kandula, MD, Dev Chethan, MD.
CARE HOSPITAL, BANJARA HILLS, Hyderabad, India.

MON-062
Background
PORD (P450 oxidoreductase deficiency) is a rare form of CAH with marked phenotypic variations due to differences in the degree of steroid hormone excess/deficiency. PORD results in 17α-hydroxylase/17,20lyase- CYP17, 21hydroxylase- CYP21, and aromatase- CYP19A1 inhibition. In the absence of characteristic skeletal features of Antley Bixler phenotype, differentiating PORD from other types of CAH is challenging.

Case details
04 day child, second of the non-identical twins, product of 2nd degree consanguinity, third in birth order was brought with abnormal genitalia. The other male twin had no genital ambiguity but had pigmented scrotum. There was no adrenal crisis in index case or maternal virilization. First child is normal female. Child was hemodynamically stable weighed 02 kgs and measured 51 cm, had no hyperpigmentation, skeletal deformities or dysmorphic features. Phallus was 10mm, clitoral index 40mm² with single urogenital opening and posteriorly fused labia (anogenital ratio 0.6). Gonads were not palpable. Karyotype was 46XX with normal Mullerian structures and non-visualized gonads on ultrasonography. Biochemical workup showed random plasma glucose level of 99mg/dl and normal electrolytes. Baseline serum 8am Cortisol was 1.15 mcg/dl (normal 5-18 mcg/dl) and 170HP was 20 ng/ml (normal < 02 ng/ml).
Serum Androstenedione level was 0.39 nmol/L (normal 1-4.8 nmol/L).

46 XX DSD Due to POR Deficiency
Nithin Modhugu Reddy, MD, Bipin Kumar Sethi, MD,DM.
Srinivas G.N.S.V. Kandula, MD, Dev Chethan, MD.
CARE HOSPITAL, BANJARA HILLS, Hyderabad, India.

MON-062
Background
PORD (P450 oxidoreductase deficiency) is a rare form of CAH with marked phenotypic variations due to differences in the degree of steroid hormone excess/deficiency. PORD results in 17α-hydroxylase/17,20lyase- CYP17, 21hydroxylase- CYP21, and aromatase- CYP19A1 inhibition. In the absence of characteristic skeletal features of Antley Bixler phenotype, differentiating PORD from other types of CAH is challenging.

Case details
04 day child, second of the non-identical twins, product of 2nd degree consanguinity, third in birth order was brought with abnormal genitalia. The other male twin had no genital ambiguity but had pigmented scrotum. There was no adrenal crisis in index case or maternal virilization. First child is normal female. Child was hemodynamically stable weighed 02 kgs and measured 51 cm, had no hyperpigmentation, skeletal deformities or dysmorphic features. Phallus was 10mm, clitoral index 40mm² with single urogenital opening and posteriorly fused labia (anogenital ratio 0.6). Gonads were not palpable. Karyotype was 46XX with normal Mullerian structures and non-visualized gonads on ultrasonography. Biochemical workup showed random plasma glucose level of 99mg/dl and normal electrolytes. Baseline serum 8am Cortisol was 1.15 mcg/dl (normal 5-18 mcg/dl) and 170HP was 20 ng/ml (normal < 02 ng/ml).
Serum Androstenedione level was 0.39 nmol/L (normal 0.5-3.4 nmol/L).