SUN-597
Prader-Willi syndrome (PWS) is a complex orphan endocrine disease characterized by hyperphagia and abnormal food-related behaviors that contribute to severe morbidity and early mortality along with a significant burden on patients and caregivers. Life-long medical care is required but the consistency of services rendered to this population has not been evaluated. This study characterized use of US hospital care, specialty physician care, and growth hormone (GH) therapy for PWS patients at different life stages.

Methods: PWS ICD-9 codes in the IQVIA™ Health Plan Claims Data from 1/2006 to 9/2015 were used to identify PWS patients. Inclusion criteria considered patients <65 years of age with ≥2 months of continuous enrollment who received ≥2 PWS diagnostic codes. Observation time was segmented into 12-month patient-years for analysis. Standardized billing code conventions were used to identify and categorize services of interest from 1/2006 to 11/2018.

Results: A total of 5,060 PWS patient years representing 1,461 unique patients were eligible. Mean annual visits to inpatient, emergency department, and physician office settings ranged by age-cohort in years from 0.2-1.6, 0.5-1.3, and 9.2-26.0, respectively. Younger (0-17) and older (50-64) age-cohorts utilized more services than early-mid adulthood age-cohorts. Use of pediatricians or endocrinologists ranged from 76% to 88% among patients under 18 years of age. Utilization of cardiologists, orthopedists, physical therapists, and otolaryngologists ranged by age-cohort from 8-44%, 7-21%, 3-21%, and 7-38%, respectively, with highest utilization among younger patients. GH use increased from 37% to 46% of PWS patients between 2007 and 2018. GH users <18 years of age were 3.0, 0.6, 1.9, 1.7, and 1.4 times as likely to use endocrinologists, cardiologists, orthopedists, physical therapists, and otolaryngologists, respectively, compared with non-GH users.

Conclusions: Use of hospital services for PWS patients was bimodal with higher use among the youngest and oldest age-cohorts. Change in the utilization level of select specialists reflects the complexity of care for age-related clinical sequelae, such as orthopedic concerns in infancy and early-onset cardiovascular disease due to hyperphagia and obesity in adolescents, as well as syndrome-specific treatment protocols (e.g., specialty consults needed for GH treatment). That less than half of PWS patients <18 years of age received GH therapy despite growing clinical evidence on the benefits and tolerability of GH suggests a potential gap in provider knowledge of the standard of care for PWS.

Our analysis suggests that GH use may be a surrogate for better access to a multidisciplinary care team and specialty services. Considerable variation of services indicates that more effort is required to optimize care in PWS.

Tumor Biology
ENDOCRINE NEOPLASIA CASE REPORTS I

Immunotherapy Use in Adrenocortical Carcinoma with Encouraging Results - a Case Report.
Parvathy Madhavan, MBBS1, Steven Shichman, MD2, Nitya Raj, MD2, Diane L. Reidy-Lagunes, MD1, Carl D. Malchoff, MD, PHD1.

Adrenal
ADRENAL CASE REPORTS I

Metastatic Paraganglioma Secondary to SDHB Gene Mutation: A Case Report and Review of New Therapies
Maryam Tetlay, M.D.1, Farhad Hasan, MD2.
A 54 years old male presented to the Emergency Department with severe left lower quadrant pain. He additionally reported recent episodes of headaches and uncontrolled hypertension, as well as an approximate weight loss of 50 pounds in the preceding two years.

A CT abdomen and pelvis demonstrated an 8 cm left peri-aortic retroperitoneal mass, along with a 2 cm solitary hepatic lesion concerning for metastatic disease. Normetanephrine levels were elevated at 1210 pg/mL (normal 0-145 pg/mL). Endoscope-guided biopsy of the retroperitoneal mass demonstrated findings consistent with a PGL.

The patient was referred to a genetic counsellor where he underwent PGLNext testing. He tested positive for a c.418G>T mutation in the SDHB gene, consistent with a diagnosis of hereditary paraganglioma - pheochromocytoma (PGL-Pheo) syndrome.

Patient was planned for surgical debulking of his PGL, and was treated pre-operatively with Phenoxybenzamine, followed by B-blockers. He underwent resection of the retroperitoneal mass and wedge resection of the hepatic mass. Surgical pathology revealed a 10.5 cm extra adrenal PGL and a 2 cm metastatic PGL involving the liver with negative margins of resection.

Patient did well in the post-operative period, and was discharged with plans to repeat plasma metanephrines in 6-8 weeks. The patient was referred to the National Cancer Institute (NCI) for enrollment in a clinical trial using Lu-177-DOTATATE. Patient did well in the post-operative period, and was discharged with plans to repeat plasma metanephrines in 6-8 weeks. The patient was referred to the National Cancer Institute (NCI) for enrollment in a clinical trial using Lu-177-DOTATATE.

Conclusion: Though rare, pheochromocytomas (pheo) and pheochromocytomas (pheo) and PGL can be lethal if not diagnosed and managed appropriately. As of 2019, at least 19 genes have been identified that predispose to familial pheochromocytoma/PGL. Importantly, PGL that are associated with SDHB, the mutation our patient had, are more likely to appear in the abdomen (rather than the head and neck), are usually secretory (most commonly norepinephrine), and are more likely to become metastatic (21-79%). Patients with metastatic pheo/PGL require complex care in specialized centers by a multidisciplinary team of specialists. Patients with advanced or non-resectable metastatic pheo/PGL should preferably be offered a referral to NCI or an NCI designated cancer center for enrollment in a clinical trial of targeted therapies.

SAT-186
Metastatic Paraganglioma Secondary to SDHB gene mutation: A Case Report and Review of New Therapies

Background: Paragangliomas (PGL) are rare neuroendocrine tumors that arise from the extra-adrenal autonomic paraganglia, majority of which are benign. 0-36% of PGL patients can develop metastatic disease, depending on the genetic mutation. We present a case of a patient with metastatic PGL due to an SDHB gene mutation. We then summarize published and ongoing trials of therapies for metastatic pheochromocytoma/PGL.

Clinical Case: A 54 years old male presented to the Emergency Department with severe left lower quadrant pain. He additionally reported recent episodes of headaches and uncontrolled hypertension, as well as an approximate weight loss of 50 pounds in the preceding two years.

A CT abdomen and pelvis demonstrated an 8 cm left peri-aortic retroperitoneal mass, along with a 2 cm solitary hepatic lesion concerning for metastatic disease. Normetanephrine levels were elevated at 1210 pg/mL (normal 0-145 pg/mL). Endoscope-guided biopsy of the retroperitoneal mass demonstrated findings consistent with a PGL.

The patient was referred to a genetic counsellor where he underwent PGLNext testing. He tested positive for a c.418G>T mutation in the SDHB gene, consistent with a diagnosis of hereditary paraganglioma - pheochromocytoma (PGL-Pheo) syndrome.

Patient was planned for surgical debulking of his PGL, and was treated pre-operatively with Phenoxybenzamine, followed by B-blockers. He underwent resection of the retroperitoneal mass and wedge resection of the hepatic mass. Surgical pathology revealed a 10.5 cm extra adrenal PGL and a 2 cm metastatic PGL involving the liver with negative margins of resection.

Patient did well in the post-operative period, and was discharged with plans to repeat plasma metanephrines in 6-8 weeks. The patient was referred to the National Cancer Institute (NCI) for enrollment in a clinical trial using Lu-177-DOTATATE.

Conclusion: Though rare, pheochromocytomas (pheo) and paragangliomas due to genetic syndromes or known germline mutations need ongoing clinical follow and biochemical testing for timely case detection. While in NF-1 pheochromocytomas are usually unilateral and present during early adulthood, they can present with bilateral disease and later in life. During surgical treatment, even with unilateral disease, adrenocortical function should be preserved when possible. Hypertension may persist despite complete surgical resection.

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