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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 ≥12 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 utilize 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.

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SUN-911

Adrenocortical carcinoma (ACC) is a rare cancer with a very poor prognosis with median survival of around 17 months. We present a patient with metastatic ACC in whom the response to PD1 inhibitor pembrolizumab has been promising. Case report: A Caucasian female patient presented at the age of 19 y with weight gain, hypertension, moon facies, supraclavicular fullness and increased hair growth on her upper back. Serum potassium concentration was 2.6 mmol/l, cortisol was 46 mcg/dL (nl = 3-12mcg/dL), and plasma ACTH concentration was < 5 pg/ml (nl = 6-58pg/ml). CT abdomen revealed a 5.4 cm right adrenal mass. For ACTH-independent Cushing's Syndrome, she underwent right adrenalectomy within 1 week of presentation. Pathology revealed a 6.5 cm ACC with negative margins, with sinusoidal invasion, but no vascular or capsular invasion. Mitosis rate was 15-20/HPF with atypical mitotic figures. Immunohistochemistry showed no loss of expression of mismatch repair gene products associated with microsatellite instability. She was unable to tolerate mitotane. Genetic analysis was negative for TP53 mutation, and she underwent radiation to the adrenal bed within 6 months following adrenalectomy. She remained without biochemical or structural evidence of disease recurrence until 2.5 years following adrenalectomy, when AM cortisol was 6 mcg/dL (nl < 1.8) after 1 mg and after 2 mg of dexamethasone the previous evening. CT scan of the pelvis, abdomen, and chest revealed 5 solid masses scattered within the lungs. The largest of these being 2.3 cm and 2 cm, and the other 3 being approximately 1 cm. Fine needle aspiration biopsy of the lung lesion revealed ACC metastases. Immunotherapy with pembrolizumab 200 mg every 3 weeks was initiated and continued for 2 years, with a side effect being grade 1 diarrhea. At 1 year after initiating pembrolizumab, she developed primary adrenal insufficiency that is being treated with 0.1 mg/d of fludrocortisone and low dose glucocorticoid replacement (hydrocortisone: 10mg in the morning and 5 mg in the evening), to avoid immune suppression. Pulmonary nodules decreased in size to 6 mm over the 2 years of pembrolizumab therapy and remained stable in size 1 year following completion of pembrolizumab therapy at which time the early morning serum cortisol concentration remained undetectable with a plasma ACTH concentration of 1177 pg/ml (nl = 6-50 pg/ml). In summary, this patient with ACC with normal mismatch repair gene expression demonstrated both structural and biochemical responses to 2 years of pembrolizumab therapy. The major side effect has been primary adrenal insufficiency. The biochemical and structural responses have been durable for 1 year after completion of pembrolizumab therapy.

Conclusion: This patient with microsatellite stable ACC has had a 36-month response to pembrolizumab.

Adrenal

ADRENAL CASE REPORTS I

Metastatic Paraganglioma Secondary to SDHB Gene Mutation: A Case Report and Review of New Therapies

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