### Beckwith-Wiedemann Syndrome

**Inheritance:** 20q13.2; Potential Fetal growth abnormalities, macroglossia, hypoglycemia, tumors and, rarely, cardiomyopathy.

**Prevalent Diagnoses:**
- CDC/Mammalian: U/S and measurement of maternal serum AFP

**Clinical Features:**
- Renal anomalies: medullary dysplasia, duplicated collecting system, nephrocalcinosis, medullary sponge kidney, cystic changes, nephromegaly
- Lab Data:
  - Airway sufficiency and feeding ability in presence of macrocycgia
  - CBF evaluation, comprehensive cardiac evaluation

**Management:**
- Referral to pediatric surgeon, psychologist, psychiatrist, endocrinologist

### Von Hippel-Lindau Syndrome

**Inheritance:** 3p25-27.3, 9p21.3, 11p15.1, 11p15.4, 11q23.1, 12q13.11 (MYH2T2) 12q13.3-q21 (FHIT3) 7q14 (HHT5)

**Prevalent Diagnoses:**
- CDC/Mammalian: Pancreatic involvement; Hepatic involvement

**Clinical Features:**
- Retinal anomalies: retinal capillary malformation, and neuroendocrine tumors of the pancreas
- Papillary cystadenomas of the epididymis and broad ligament
- Lab Data:
  - clinical signs of ELH; measurement of urinary catecholamine metabolites, urine; rules out genetic pathologies

**Management:**
- Referral to geneticist, ENT, nephrologist/urologist, endocrinologist

### Cutis Marmorata Telangiectatica Congenita

**Inheritance:** 9p13-4.13 (cyc1 D1 gene)

**Prevalent Diagnoses:**
- Endothelial sac tumors of the middle ear
- Cystic kidney and renal abnormalities
- Papillary cystadenomas of the epididymis and broad ligament
- Lab Data:
  - clinical signs of ELH; measurement of urinary catecholamine metabolites, urine; rules out genetic pathologies

**Management:**
- Referral to geneticist, ENT, nephrologist/urologist, endocrinologist

### Maffucci Syndrome

**Inheritance:** Genetic

**Prevalent Diagnoses:**
- Congenital/developmental abnormalities
- Neurocutaneous abnormalities

**Clinical Features:**
- Neuroectodermal abnormalities
- KTS, Nevox Anemosis

**Management:**
- Empirical treatment
- Referral to orthopedist and/or neurosurgeon

### Kasabach-Merritt Syndrome

**Inheritance:** Genetic

**Prevalent Diagnoses:**
- Erythromelalgia by thrombocytopenia (PTHR1)
- Associated somatic mutations in the kis3tide dehydrogenase-1 and -2 genes

**Clinical Features:**
- Physical exam
- Evaluation of leukemia

**Management:**
- Referral to hematology

### Blue Rubber Bleb Nevus Syndrome

**Inheritance:** 3p25-27.3, 9p21.3, 11p15.1, 11p15.4, 11q23.1, 12q13.11 (MYH2T2) 12q13.3-q21 (FHIT3) 7q14 (HHT5)

**Prevalent Diagnoses:**
- CDC/Mammalian: Pancreatic involvement; Hepatic involvement

**Clinical Features:**
- Retinal anomalies: retinal capillary malformation, and neuroendocrine tumors of the pancreas
- Papillary cystadenomas of the epididymis and broad ligament
- Lab Data:
  - clinical signs of ELH; measurement of urinary catecholamine metabolites, urine; rules out genetic pathologies

**Management:**
- Referral to geneticist, ENT, nephrologist/urologist, endocrinologist

### Hereditary Hemorrhagic Telangiectasia Syndrome

**Inheritance:** 1q23.3; 11p15.1, 12q13.11 (MYH2T2) 12q13.3-q21 (FHIT3) 7q14 (HHT5)

**Prevalent Diagnoses:**
- CDC/Mammalian: Pancreatic involvement; Hepatic involvement

**Clinical Features:**
- Retinal anomalies: retinal capillary malformation, and neuroendocrine tumors of the pancreas
- Papillary cystadenomas of the epididymis and broad ligament
- Lab Data:
  - clinical signs of ELH; measurement of urinary catecholamine metabolites, urine; rules out genetic pathologies

**Management:**
- Referral to geneticist, ENT, nephrologist/urologist, endocrinologist

### Proteus Syndrome

**Inheritance:** Genetic

**Prevalent Diagnoses:**
- Erythromelalgia by thrombocytopenia (PTHR1)
- Associated somatic mutations in the kis3tide dehydrogenase-1 and -2 genes

**Clinical Features:**
- Physical exam
- Evaluation of leukemia

**Management:**
- Referral to hematology

### PHACE Syndrome

**Pathogenesis:**
- Timing of morphogenesis errors occur between 3-12 weeks of gestation
- Many elements of disease could be due to abnormality of cell proliferation and apoptosis

**Key Features:**
- Association of CNS abnormalities with developmental delays, motor delays and pterygium dysplasia
- Endocrinologic abnormalities reported in a small number of patients
- Sternal clefting/supramammillary raphé
- Association of hearing loss

**Management:**
- Referral to neurologist

### Bibliography