Beckwith-Wiedemann Syndrome

Main Features

- Pediatric overgrowth syndrome
  - height and weight >97% in first few years of life
  - Hemihyperplasia
  - Macroglossia
  - Abdominal wall defects (omphalocele, umbilical hernia, diastasis recti)
  - Visceromegaly
    - single organs or in combination (liver, spleen, pancreas, kidneys, adrenals)
    - Fetal adrenocortical cytomegaly
  - Predisposition to embryonal tumor development 7.5%
    - Most occur in the first 8-10 years of life
    - Wilms tumor and hepatoblastoma most common
    - rhabodomyosarcoma
    - adrenocortical carcinoma
    - neuroblastoma

Eye Findings

- Strabismus
- Hypertelorism, exophthalmos
- Nystagmus
- Infraorbital creases

Other Findings

- Cardiac malformations 20%
- Renal malformations (unilateral or bilateral)
  - Malformation, renal medullary dysplasia, nephrocalcinosis, nephrolithiasis
- Nevus flammeus
- mid facial hypoplasia
- full lower face with prominent mandible

Etiology

- Chromosome 11p15.5 abnormality
  - Hypomethylation of differentially methylated region 2 (DMR2)
  - gain of methylation of differential methylated region 1 (DMR1)
  - CDKN1C mutations
- Increased risk for children born with assistive reproductive techniques

- estimated incidence 1:13,700

Reference

- Beckwith-Wiedeman syndrome. Weksberg, Shuman and Beckwith. European Journal of Medical Genetics 2010
- Ocular manifestations of Beckwith-Wiedemann sydrome. Rohewetz et al. JAAPOS 2021

syndrome