

An **overgrowth disorder** resulting from epigenetic or genetic alterations, such as **abnormal methylation** or a pathogenic variant in CDKN1C (Cyclin-Dependent Kinase Inhibitor 1C). It follows an **autosomal dominant** inheritance pattern, though most cases are **sporadic**.

AD
Autosomal dominant

CLINICAL FEATURES

Growth:

- **Macrosomia**
- **Hemihypertrophy**
- Rapid growth
- Growth rate slows after 8 years of age



Facial features:

- **Macroglossia**
- **Earlobe creases**
- Earlobe pits
- Nevus flammeus
- Infraorbital creases



Gastrointestinal:

- **Omphalocele**
- **Umbilical hernia**
- Visceromegaly



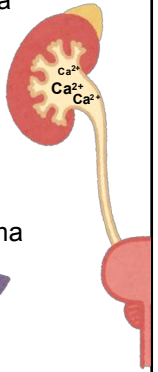
Endocrine:

- **Hypoglycemia**
- Persistent hyperinsulinism



Renal:

- Medullary dysplasia
- **Nephrocalcinosis**



Embryonal tumors:

- **Wilms tumor**
- **Hepatoblastoma**
- Neuroblastoma
- Rhabdomyosarcoma



DIAGNOSIS

- Identification of **key clinical features**

Confirmatory Genetic Testing:

- **DNA methylation studies**
- **Chromosomal microarray** to confirm microdeletions/duplications, or paternal uniparental disomy



CANCER SCREENING

Early diagnosis and regular cancer screening are essential.

- **Abdominal ultrasound**
 - Every 3 months until 8 years old
- **Alpha-fetoprotein level (AFP) levels**
 - Every 3 months until 4 years old
- **Renal bladder ultrasound**
 - Annually from 8 years to adolescence



MANAGEMENT

- **Genetic counseling:** to understand genetic risks and implications.
- **Developmental monitoring:** track milestones and address any delays.
- **Macroglossia:** affects ~ 90% of children with BWS.
 - Feeding support and/or surgical management.
- **Abdominal wall defects:** may require surgical repair.
- **Hypoglycemia management:** treatment may involve diazoxide, octreotide, continuous feeds, or partial pancreatectomy.
- **Leg-length differences:** orthotics or other methods can be used to address differences in leg length.



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