

A 4-month-old boy is brought to the physician for routine evaluation. He spent 3 weeks in the neonatal intensive care unit for management of hypoglycemia and poor feeding secondary to macroglossia. The patient has had no significant issues since discharge home. He is currently at the 99th percentile for weight, length, and head circumference. Physical examination shows an enlarged tongue and a reducible umbilical hernia. His right upper and lower extremities are significantly larger in circumference than the left extremities. The remainder of his examination is normal. In addition to routine vaccinations, which of the following is the best next step in management of this patient?

- ☐ A. Abdominal ultrasound
- ☐ B. Brain magnetic resonance imaging
- ☐ C. Referral for herniorrhaphy
- ☐ D. Serum glucose level
- ☐ E. Thyroid-stimulating hormone level
- ☐ F. Urine homovanillic and vanilmandelic acid

Submit

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- ☒ A. Abdominal ultrasound [35%]
- ☐ B. Brain magnetic resonance imaging [9%]
- ☐ C. Referral for herniorrhaphy [1%]
- ☐ D. Serum glucose level [10%]
- ☐ E. Thyroid-stimulating hormone level [41%]
- ☐ F. Urine homovanillic and vanilmandelic acid [4%]

Proceed to Next Item

Explanation:

User Id:

Beckwith-Wiedemann syndrome	
Pathogenesis	<ul style="list-style-type: none">• Deregulation of imprinted gene expression in chromosome 11p15
Physical examination	<ul style="list-style-type: none">• Fetal macrosomia, rapid growth until late childhood• Omphalocele or umbilical hernia• Macroglossia• Hemihyperplasia
	<ul style="list-style-type: none">• Wilms tumor

Explanation:

User Id: [REDACTED]

Beckwith-Wiedemann syndrome	
Pathogenesis	<ul style="list-style-type: none"> Deregulation of imprinted gene expression in chromosome 11p15
Physical examination	<ul style="list-style-type: none"> Fetal macrosomia, rapid growth until late childhood Omphalocele or umbilical hernia Macroglossia Hemihyperplasia
Complications	<ul style="list-style-type: none"> Wilms tumor Hepatoblastoma
Surveillance	<ul style="list-style-type: none"> Serum alpha fetoprotein Abdominal/renal ultrasound

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Beckwith-Wiedemann syndrome (BWS) is an overgrowth disorder characterized by a predisposition to neoplasms. Most patients have a sporadic or inherited alteration of chromosome 11p15, which includes genes that encode insulin-like growth factor 2, a growth-promoting hormone similar to insulin. At birth, classic physical findings include **macrosomia**, **macroglossia**, **hemihyperplasia**, and medial abdominal wall defects (**umbilical hernia**, **omphalocele**). Some infants also have visceromegaly.

Newborns must be monitored closely for **hypoglycemia**. Fetal hyperinsulinemia can result in profound hypoglycemia at birth (similar to infants of diabetic mothers). This problem is usually transient, and older asymptomatic patients usually do not require ongoing glucose monitoring (**Choice D**). Patients with BWS are at significantly increased risk of **Wilms tumor** and **hepatoblastoma**. Screening **abdominal ultrasound** and **α-fetoprotein** levels should occur every 3 months from birth to age 4

Surveillance

- Serum alpha fetoprotein
- Abdominal/renal ultrasound

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Patients with isolated hemihyperplasia are also at increased risk for Wilms tumor and hepatoblastoma. These patients should undergo frequent screening as in BWS.

(Choice B) Brain magnetic resonance imaging should be performed in patients with stigmata of neurocutaneous syndromes (eg, neurofibromatosis, tuberous sclerosis, von Hippel-Lindau syndrome). Although patients with BWS are at increased risk for various abdominal neoplasms, brain lesions are not typical features of this disease.

(Choice C) Many umbilical hernias close in the first few years of life. In contrast to inguinal hernias, umbilical hernias are seldom symptomatic and are at much lower risk of incarceration/strangulation. Therefore, repair is rarely recommended until age 5 years.

(Choice E) Congenital hypothyroidism also can present with macroglossia and umbilical hernia. However, macrosomia, hypoglycemia, and hemihyperplasia are not features of congenital hypothyroidism, and this endocrinopathy is not associated with BWS.

(Choice F) Patients with BWS are at somewhat increased risk of neuroblastoma. However, routine screening with urinary homovanillic and vanilmandelic acid assays is not recommended due to the low incidence of this associated tumor.

Educational objective:

Beckwith-Wiedemann syndrome is characterized by macrosomia, macroglossia, umbilical hernia/omphalocele, hemihyperplasia, and hypoglycemia. Children must be closely monitored for development of Wilms tumor or hepatoblastoma.

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References:

1. [Experience with hemihyperplasia and Beckwith-Wiedemann syndrome surveillance protocol.](#)
2. [Tumour surveillance in Beckwith-Wiedemann syndrome and hemihyperplasia: a critical review of the evidence and suggested guidelines for local practice.](#)
3. [Serum alpha-fetoprotein screening for hepatoblastoma in children with Beckwith-Wiedemann syndrome or isolated hemihyperplasia.](#)
4. [Beckwith-Wiedemann syndrome: historical, clinicopathological, and etiopathogenetic perspectives.](#)

Media Exhibit

hernia



