

Question 18 – Week of November 1

Which of these does not characterize infants with Beckwith-Wiedemann syndrome?

- A. Hypoglycemia
- B. Mental retardation
- C. Atrophic gastritis
- D. Omphalocele
- E. Trisomy 13,18

Answer: C

Infants with the Beckwith-Wiedemann syndrome (mental retardation, hepatomegaly, large body stature, hypoglycemia in early infancy) may be born with omphalocele which involves a herniation of the umbilicus. Three of four patients with omphalocele have extraintestinal birth defects, most commonly trisomy 13 or 18. Increased levels of maternal serum alpha-fetoprotein suggest the presence of possible omphalocele. Ultrasound examination during pregnancy allows the diagnosis of this abdominal wall defect in most infants.

Reference:

Feldman: Sleisenger & Fordtran's Gastrointestinal and Liver Disease, 8th ed.