

## SPOT DIAGNOSIS (IMAGE GALLERY)



### OVERWEIGHT BABY

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A 2 month old overweight baby (weight 6.3 kg >95th percentile and height 59cm at 50th percentile) born of non consanguineous marriage came with history of hypoglycemic convulsions since day 15 of life. On examination, child had microcephaly (32 cm <3 centile), umbilical hernia and a large tongue.

### What is the diagnosis?

Beckwith-Wiedemann syndrome. John Bruce Beckwith, an American pathologist and Hans-Rudolf Wiedemann, a German pediatrician, independently reported cases of a new syndrome at first. (1,2) Originally, it was termed EMG syndrome (for exomphalos, macroglossia, and gigantism) which over time came to be known as Beckwith-Wiedemann syndrome. This syndrome is characterized by macroglossia, microcephaly, exomphalos, macrosomia and visceromegaly. Distinctive lateral earlobe fissures may be present, and hemihypertrophy occurs in many of these infants. Hypoglycemia associated with hyperinsulinemia is seen in approximately 50 percent of patients. Diffuse islet cell hyperplasia occurs in infants with hypoglycemia. They may develop embryonal tumors like Wilm's tumor, hepatoblastoma, adrenal carcinoma and rhabdomyosarcoma. (3) Renal abnormalities like medullary dysplasia, nephrocalcinosis, medullary sponge kidney and nephromegaly also can occur. Growth rate slows around age seven to eight years. Hemihyperplasia may affect segmental regions of the body or selected organs and tissues. (4) This overgrowth syndrome is caused by mutations in the chromosome 11p15.5 region close to the genes for insulin, SUR, KIR6.2, and IGF-2. Duplications in this region and genetic imprinting from a defective or absent copy of the maternally derived gene are involved in the variable features and patterns of transmission. (3) Diagnosis of Beckwith-Wiedemann syndrome is primarily made by the characteristic clinical findings. Confirmation is done by genetic testing with Cytogenetic tests like FISH-metaphase 2, FISH-interphase 2 and Molecular genetic tests like methylation analysis, uniparental disomy study, deletion, duplication analysis, sequence analysis of entire coding region, web addressage analysis, targeted variant analysis. (4,5) The absence of a mutation in a child with clinical findings suggestive should not preclude a diagnosis of this disease. Management is done by preventing hypoglycemia by frequent feeds and drugs like diazoxide. Hypoglycemia may resolve over weeks to months of medical therapy. Pancreatic resection may also be needed. (3) Abdominal wall defect is treated by surgical correction at the earliest. Feeding problems due to macroglossia can be corrected by tongue reductive surgery. Patients should be screened routinely for malignancies and most of them are treatable by surgery or chemotherapy. Prognostically, most of the children who survive infancy do well, the older the child becomes, more normal they appear. IQ may be normal or low if child has got hypoglycemic insult. Prenatal screening for pregnant mothers by ultrasound examination and maternal serum alpha-fetoprotein assay may help to consider chromosomal analysis and, or molecular genetic testing in suspected fetuses which is done especially in families having history of this disease. (4)

### References

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