Beckwith-Wiedemann Syndrome

Alternative Names
BWS
EMG Syndrome
Exomphalos-Macroglossia-Gigantism Syndrome
Wiedemann-Beckwith Syndrome
WBS
Beckwith-Wiedemann Syndrome Chromosome Region
BWCR

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Other congenital malformations

Incidence per 100,000 Live Births
6-10

OMIM Number
130650

Mode of Inheritance
Autosomal dominant

Gene Map Locus
11p15.5, 11p15.5, 11p15.5, 5q35

Description
Beckwith-Wiedemann Syndrome (BWS) is a congenital growth disorder characterized by the EMG triad; Exomphalos-Macroglossia-Gigantism. Infants are born large for their gestational age, have a large tongue, prominent eyes, abnormal low-set ears, and creases in the ear lobes. Other abnormalities seen include hypoglycemia, defects in the abdominal wall (umbilical hernia, diastasis recti, omphalocele), cryptorchidism, renal abnormalities, hemihypertrophy, seizures, poor feeding, and lethargy. Infants are at an increased rate of developing tumors, the most common being Wilm's tumor and adrenal carcinoma. Worldwide, one in approximately every 13,700 births is estimated to be affected by BWS. Interestingly, children conceived by *in vitro* fertilization have been noticed to be three to four times more likely to develop this condition.

BWS is suspected in large sized infants with hypoglycemia, organ enlargement, and enlarged fontanelle. Diagnosis can be confirmed with the help of X-ray of bones, MRI or CT scan of the abdomen, and chromosomal studies, especially in chromosome 11. Treatment of the condition involves multiple strategies. The hypoglycemia needs to be treated in infancy with intravenous glucose infusions as well as hydrocotisone. Tongue size can be corrected with surgery. As infants grow up, the complications of BWS reduce dramatically. Children need to be monitored for tumor development, although the risk decreases significantly after around 7-years of age.

Molecular Genetics
The genetic basis of BWS is considered to be heterogeneous. The locus 11p15 is considered to be the most important for this condition, with at least three different regions in this loci being implicated in the development of BWS. Incidentally, this locus was the first example of imprinting in mammals, and not surprisingly, up to 20% of cases of BWS show uniparental disomy of 11p15. Of the genes in this locus which have been implicated in the development of BWS, the most important ones are H19, IGF2 (Insulin Like Growth Factor 2), CDKN1C (Cyclin Dependent Kinase Inhibitor 1C), and ZNF215 (Zinc Finger Protein 215).

Epidemiology in the Arab World
Mauritania
Ndiaye et al. (2006) report the case of a two month old child presenting with hemihypertrophy, macroglassia and an umbilical hernia. Glycemia was under normal level showing a mild hypoglycemia (0.6

g/dl). T3, T4 and TSH values were in normal range. Abdominal echography was normal.

Oman

Sawardekar (2005) conducted a study to establish the prevalence of major congenital malformations in children born during a 10-year period in Nizwa Hospital. Of the 21,988 total births in the hospital, two children were born with Beckwith-Wiedemann Syndrome.

References

Ndiaye O, Diouf S, Fall AL, Sylla A, Guèye M, Ouattara A, Sall MG, Kuakuvi N. [Beckwith-Wiedemann syndrom: a case report in Dakar] Dakar Med. 2006; 51(2):101-3. PMID: 17632986

Sawardekar KP. Profile of major congenital malformations at Nizwa Hospital, Oman: 10-year review. J Paediatr Child Health. 2005; 41(7):323-30. PMID: 16014135

Related CTGA Records

N/A

External Links

http://atlasgeneticsoncology.org/Kprones/BeckwithWiedemannID10037.html

http://www.beckwith-wiedemannsyndrome.org/tp42/Default.asp?ID=28722

http://www.bws-support.org.uk/

http://www.emedicine.com/PED/topic218.htm

http://www.genetests.org/profiles/bws

http://www.nlm.nih.gov/medlineplus/ency/article/001186.htm

http://www.orpha.net/data/patho/GB/uk-BWS05.pdf

Contributors

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