Benign familial neonatal seizures

Benign familial neonatal seizures (BFNS) is a condition characterized by recurrent seizures in newborn babies. The seizures begin around day 3 of life and usually go away within 1 to 4 months. The seizures can involve only one side of the brain (focal seizures) or both sides (generalized seizures). Many infants with this condition have generalized tonic-clonic seizures (also known as grand mal seizures). This type of seizure involves both sides of the brain and affects the entire body, causing muscle rigidity, convulsions, and loss of consciousness.

A test called an electroencephalogram (EEG) is used to measure the electrical activity of the brain. Abnormalities on an EEG test, measured during no seizure activity, can indicate a risk for seizures. However, infants with BFNS usually have normal EEG readings. In some affected individuals, the EEG shows a specific abnormality called the theta pointu alternant pattern. By age 2, most affected individuals who had EEG abnormalities have a normal EEG reading.

Typically, seizures are the only symptom of BFNS, and most people with this condition develop normally. However, some affected individuals develop intellectual disability that becomes noticeable in early childhood. A small percentage of people with BFNS also have a condition called myokymia, which is an involuntary rippling movement of the muscles. In addition, in about 15 percent of people with BFNS, recurrent seizures (epilepsy) will come back later in life after the seizures associated with BFNS have gone away. The age that epilepsy begins is variable.

Frequency

Benign familial neonatal seizures occurs in approximately 1 in 100,000 newborns.

Causes

Mutations in two genes, \( \text{KCNQ2} \) and \( \text{KCNQ3} \), have been found to cause BFNS. Mutations in the \( \text{KCNQ2} \) gene are a much more common cause of the condition than mutations in the \( \text{KCNQ3} \) gene.

The \( \text{KCNQ2} \) and \( \text{KCNQ3} \) genes provide instructions for making proteins that interact to form potassium channels. Potassium channels, which transport positively charged atoms (ions) of potassium into and out of cells, play a key role in a cell’s ability to generate and transmit electrical signals.

Channels made with the KCNQ2 and KCNQ3 proteins are active in nerve cells (neurons) in the brain, where they transport potassium ions out of cells. These channels transmit a particular type of electrical signal called the M-current, which prevents the
neuron from continuing to send signals to other neurons. The M-current ensures that the neuron is not constantly active, or excitable.

Mutations in the \textit{KCNQ2} or \textit{KCNQ3} gene result in a reduced or altered M-current, which leads to excessive excitability of neurons. Seizures develop when neurons in the brain are abnormally excited. It is unclear why the seizures stop around the age of 4 months. It has been suggested that potassium channels formed from the KCNQ2 and KCNQ3 proteins play a major role in preventing excessive excitability of neurons in newborns, but other mechanisms develop during infancy.

About 70 percent of people with BFNS have a mutation in either the \textit{KCNQ2} or the \textit{KCNQ3} gene. Researchers are working to identify other gene mutations involved in this condition.

\textbf{Inheritance Pattern}

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person inherits the mutation from one affected parent. A few cases result from new mutations in the \textit{KCNQ2} gene. These cases occur in people with no history of benign familial neonatal seizures in their family.

\textbf{Other Names for This Condition}

- benign familial neonatal convulsions
- benign familial neonatal epilepsy
- benign neonatal convulsions
- benign neonatal epilepsy
- BFNS

\textbf{Diagnosis & Management}

\textbf{Genetic Testing Information}

- What is genetic testing? /primer/testing/genetictesting
Other Diagnosis and Management Resources

- Boston Children's Hospital: My Child Has...Seizures and Epilepsy
  http://www.childrenshospital.org/conditions-and-treatments/conditions/s/seizures
- Epilepsy Action: Benign Neonatal Convulsions
  https://www.epilepsy.org.uk/info/syndromes/benign-neonatal-convulsions
- GeneReview: KCNQ2-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK32534
- GeneReview: KCNQ3-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK201978
- MedlinePlus Encyclopedia: EEG
  https://medlineplus.gov/ency/article/003931.htm

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: EEG
  https://medlineplus.gov/ency/article/003931.htm
- Health Topic: Epilepsy
  https://medlineplus.gov/epilepsy.html
- Health Topic: Seizures
  https://medlineplus.gov/seizures.html

Genetic and Rare Diseases Information Center

- Convulsions benign familial neonatal dominant form

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Epilepsy
  https://www.ninds.nih.gov/Disorders/All-Disorders/Epilepsy-Information-Page

Educational Resources

- Boston Children's Hospital: My Child Has...Seizures and Epilepsy
  http://www.childrenshospital.org/conditions-and-treatments/conditions/s/seizures
- Centers for Disease Control and Prevention: Epilepsy
  https://www.cdc.gov/epilepsy/index.html
- Epilepsy Action: Benign Neonatal Convulsions
  https://www.epilepsy.org.uk/info/syndromes/benign-neonatal-convulsions
- KidsHealth from Nemours: Seizures
MalaCards: benign familial neonatal epilepsy  
https://www.malacards.org/card/benign_familial_neonatal_epilepsy

World Health Organization: Epilepsy  
https://www.who.int/en/news-room/fact-sheets/detail/epilepsy

Clinical Information from GeneReviews

- KCNQ2-Related Disorders  
  https://www.ncbi.nlm.nih.gov/books/NBK32534

- KCNQ3-Related Disorders  
  https://www.ncbi.nlm.nih.gov/books/NBK201978

Scientific Articles on PubMed

- PubMed  
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28benign+familial+neonatal+seizures%5BTIAB%5D%29+OR+%28BFNS%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SEIZURES, BENIGN FAMILIAL NEONATAL, 1  
  http://omim.org/entry/121200

- SEIZURES, BENIGN FAMILIAL NEONATAL, 2  
  http://omim.org/entry/121201

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9430594

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11784811

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16735477
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1472242/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10482260
Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20437616

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