

Homocystinuria

Cystathionine Betasynthase Deficiency, CBS Deficiency

Condition Description

Homocystinuria, an amino acid disorder, occurs when an enzyme called "cystathionine beta-synthase" (CBS) is either missing or not working properly. This enzyme's job is to break down methionine. When this enzyme is not working correctly, methionine and another amino acid, homocystine, build up in the blood and cause problems.

Range of Outcomes

There is no specific cure for homocystinuria. However, many people respond to high doses of vitamin B-6 (also known as pyridoxine). Slightly less than half of patients respond to this treatment; those that do respond need supplemental vitamin B-6 for the rest of their lives. Those who do not respond require a low methionine diet, and most will need treatment with trimethylglycine (a medication also known as betaine).

This disorder has varying effects that depend on the expression of the disorder in the individual. Without treatment, children with homocystinuria exhibit poor growth, severe developmental delay, osteoporosis, glaucoma, stroke, seizures, pancreatitis and in some cases death by age 1 year.

Minnesota now screens newborns for this rare disorder. Early treatment is complex and consists of a special dietary formula which restricts methionine and several supplements in order to minimize the effects of the disease. Lifelong dietary treatment is essential.

The relationship between the variability of the disease versus treatment compliance has not been fully determined. Infant developmental monitoring and evaluation is needed to intervene early and appropriately. A medical plan is essential and will vary depending on which problems the particular child experiences. Literature does not have extensive information about long term outcomes; there are some case reports

Common Complications

Treatment may lower the chance for blood clots, heart disease, and stroke but not eliminate it. Treatment seems to lessen the chance of eye problems; however, even when treated, more than half infants and children develop vision problems, developmental delay, and/or seizures.

If a stroke occurs, intervention will be focused on that part of the brain damaged. If a vision problem is an issue, services will need to consider this aspect. Ongoing involvement with this child and family will be the best way to determine what interventions are needed and when.

Side Effects of Treatment

The dietary and supplement treatment is complex and treatment compliance can be difficult to maintain.

For More Information

This fact sheet was developed for use in determining eligibility for early intervention services only. For more complete information, the following resources might be useful:



Minnesota Children with Special Health Needs (MCSHN)
85 E. 7th Place, Suite 220
P.O. Box 64882
St. Paul, MN 55164
1-800-728-5420 or (651) 201-3650
www.health.state.mn.us/mcshn

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MedlinePlus

MedlinePlus brings together authoritative information from NLM, the National Institutes of Health (NIH), and other government agencies and health-related organizations. Preformulated MEDLINE searches also give access to medical journals.

<http://medlineplus.gov/>

Agency for Healthcare Research and Quality

Under its Evidence-based Practice Program, the Agency for Healthcare Research and Quality (AHRQ) is developing scientific information for other agencies and organizations on which to base clinical guidelines, performance measures, and other quality improvement tools. Contractor institutions review all relevant scientific literature on assigned clinical care topics and produce evidence reports and technology assessments, conduct research on methodologies and the effectiveness of their implementation, and participate in technical assistance activities.

<http://www.ahrq.gov/clinic/epcsums/lbwdissum.htm>

E-Medicine

eMedicine's clinical knowledge base contains peer reviewed articles on a number of different health conditions. There is a pediatric section available.

<http://www.emedicine.com/>

References

Kaye, "Newborn Screening Fact Sheets" Pediatrics.2007,

Pass et al, Homocystinuria Screening in Newborns.APHL. 1993.

Mudd, "The Natural History of Homocystinuria due to CBS", Am. Jnl of Genetics, 1985.

Online Mendelian Inheritance in Man, (OMIM). www.newbornscreening.info