KENTUCKY NEWBORN SCREENING PROGRAM PARENT TEACHING SHEET

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Ornithine Transcarbamylase Deficiency (OTC)

<u>CAUSE</u>

OTC is a condition in which the body is unable to process and remove the waste, ammonia. It is considered an amino acid condition because ammonia is produced when the body breaks down proteins in food into their basic building blocks (amino acids). When the body's process for removing waste is disrupted, dangerous amounts of ammonia begin to build up in the blood. This can result in severe mental disability, seizures and avariety of other life threatening medical problems.

SYMPTOMS

Early signs of OTC include:

- Developmental delay
- Poor appetite
- Vomiting
- Sleeping longer or more often
- Irritability
- Trouble regulating body temperature
- Trouble breathing
- Involuntary or uncontrolled body movements.

TREATMENT OPTIONS

Treatment is life-long and can include low protein diet (a dietician will help you set up the best diet for your child), special formula low in protein and medications/supplements to help prevent high ammonia. Children with OTC should see their regular doctor, a doctor who specializes in OTC and a dietician.

