Patient Registry

The ACD Patient Registry allows patients, family members, and researchers to gather information in a safe, confidential, online database.

This information is critical to researchers understanding the history and progression of CCDS, to patients and families learning about treatment developments, and to advocates speaking on behalf of the CCDS community.

Please visit the registry at:
http://www.creatineinfo.org/patient-registry

Collaborative Partnerships

The Association for Creatine Deficiencies is a proud partner of the National Organization for Rare Disorders (NORD) and Global Genes Rare Foundation Alliance. Together we are part of a collaborative effort for rare diseases.

NORD National Organization for Rare Disorders Member Organization

Global Genes Allies in Rare Disease

ASSOCIATION FOR CREATINE DEFICIENCIES

EDUCATION ADVOCACY RESEARCH

The Association for Creatine Deficiencies

The Association was established to raise awareness and education of CCDS among the medical community, as well as the general public, and to advocate on behalf of families and patients living with Cerebral Creatine Deficiency Syndromes.

Together we are

BUILDING STRENGTH
changing lives

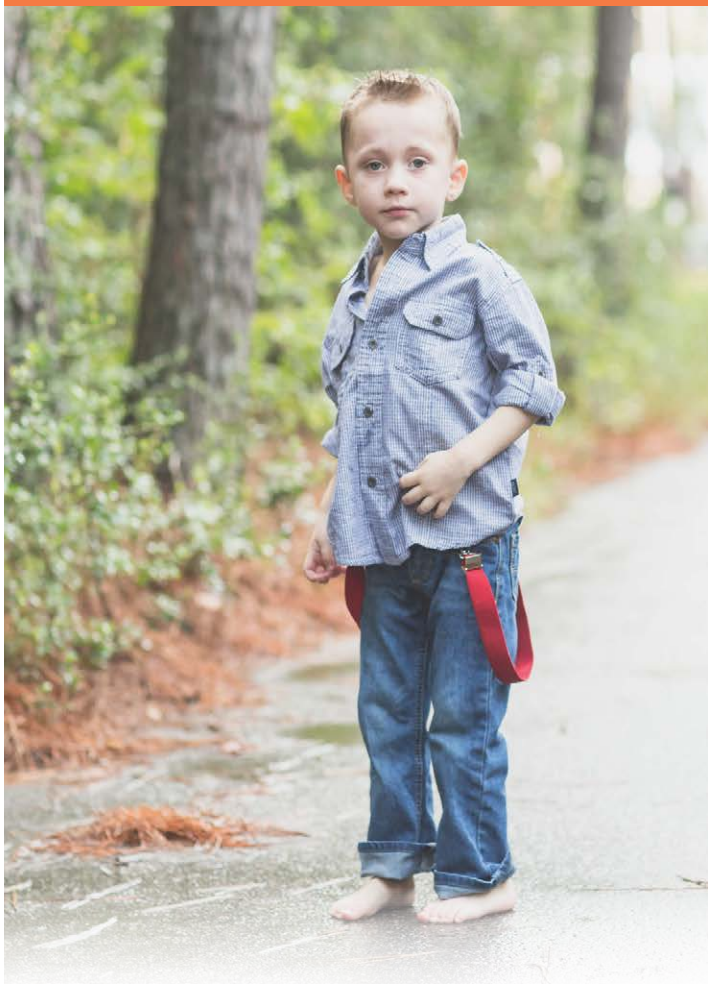
Visit Us At
www.creatineinfo.org

Contact Us At
info@creatineinfo.org

Follow us on:
/creatineinfo @creatineinfo

Guide to Understanding

Cerebral Creatine Deficiency Syndromes (CCDS)



ASSOCIATION FOR CREATINE DEFICIENCIES

EDUCATION ADVOCACY RESEARCH

Cerebral Creatine Deficiency Syndromes?

Cerebral Creatine Deficiency Syndromes are inborn errors of metabolism, which interrupt the formation or transportation of creatine. Creatine is necessary to increase adenosine triphosphate (ATP), which provides energy to all cells in the body. Creatine is essential to sustain the high energy levels needed for muscle and brain development.

There are three Cerebral Creatine Deficiency Syndromes:

Creatine Transporter Deficiency (CTD) is a mutation in the creatine transporter gene. This results in a blockage in the transportation of creatine to the brain and muscles.

Guanidinoacetate Methyltransferase Deficiency (GAMT) is a mutation in the gene that makes the enzyme that creates creatine, resulting in a shortage of creatine.

Arginine: Glycine Amidinotransferase Deficiency (AGAT) involves the first step of creatine synthesis. Mutations found in the AGAT gene impair the body's production of creatine.

What are the symptoms of CCDS?

Global developmental delay affects all children with these disorders and may be the first sign, appearing before other symptoms. Speech delay may be particularly severe and is present in all affected children. Intellectual disability of variable severity is typically present in all older children and adults.

Additional symptoms may include:

- Seizure disorders, the severity and onset are variable
- Hypotonia, muscle weakness, and muscle hypotrophy
- Behavior disorders including autism-like behaviors and hyperactivity
- Movement disorders including dystonia and dyskinesia (sometimes labeled as cerebral palsy)
- Gastrointestinal problems such as chronic constipation and vomiting
- Failure to thrive

Common CCDS Misdiagnoses

CCDS often mimic other disorders, including:

- Autism
- Cerebral Palsy
- Mitochondrial Disorder
- Failure to Thrive
- Developmental Delay and Disability

CCDS Screening

Initial screening for CCDS is non-invasive. Testing in both urine and plasma is recommended for all three deficiencies by measuring the concentration of creatine (Cr), guanidinoacetate (GAA), and creatinine (Crn). Follow up genomic testing and brain MRI with spectroscopy may be ordered to confirm CCDS diagnosis.

For information on laboratories offering testing for CCDS, go to:

GeneTests: **www.genetests.org**

The Genetic Testing Registry (GTR®): **http://www.ncbi.nlm.nih.gov/gtr/**

Treatments of CCDS

Treatment with oral supplementation is available and effective if initiated early for the AGAT and GAMT deficiencies. To date, this type of treatment has not shown to improve outcomes in individuals with CTD. The ACD does not recommend or advise any supplementation to be taken without physician supervision.