



Genetic Fact Sheets for Professionals

Amino Acid Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

Disease name	Citrullinemia
Alternate name(s)	Argininosuccinic acid synthetase deficiency
Acronym	ASAS
Disease classification	Amino Acid Disorder
Variants	Yes
Variant name	Citrullinemia type II (adult and neonatal onset forms) – caused by <i>SLC25A13</i> mutations
Symptom onset	Neonatal with some variability
Symptoms	Potential lethal coma, seizures, anorexia, vomiting, lethargy, apnea and hypertonia. Possible enlarged liver.
Natural history without treatment	Mental retardation due to hyperammonemia.
Natural history with treatment	Normal IQ and development are possible if no damage from initial or subsequent hyperammonemic episodes.
Treatment	Management of hyperammonemic cases with sodium benzoate and/or phenylacetate and arginine. Dietary restriction of protein, arginine and essential amino acid supplementation.
Other	N/A

Physical phenotype	None
Inheritance	Autosomal recessive
General population incidence	Rare
Ethnic differences	Yes
Population	Citrullinemia type II is common in Japan
Ethnic incidence	N/A
Enzyme location	Widely expressed in tissues; liver, kidney and fibroblasts.
Enzyme function	Catalyzes the conversion of citrulline and aspartic acid to argininosuccinic acid.
Missing enzyme	Argininosuccinic acid synthetase
Metabolite changes	Hyperammonemia
Gene	CTLN1
Gene location	9q34
DNA testing available	Yes
DNA testing detail	Linkage analysis
Prenatal testing	Linkage analysis and enzyme testing
MS/MS profile	N/A
OMIM link	www.ncbi.nlm.nih.gov/htbin-post/Omim/dispnim?215700
Genetests link	www.genetests.org/servlet/access?prg=j&db=genestar&site=&fcn=d&id=12600&qry=2217&res=nous&res=nointl&key=0fnnczIQk-G9&show_flag=c

Support group

National Urea Cycle Disorders Foundation
www.nucdf.org

National Coalition for PKU and Allied Disorders
www.pku-allieddisorders.org

Children Living with Inherited Metabolic Diseases
www.climb.org.uk

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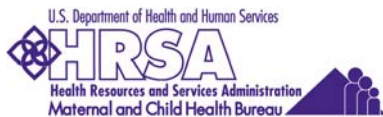
Reviewed by HI, CA, OR and WA metabolic specialists

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Updated on N/A

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