



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	Argininemia/arginase deficiency
Alternate name(s)	Hyperargininemia ARG I deficiency
Acronym	ARG I
Disease classification	Amino Acid Disorder
Variants	Yes
Variant name	ARG II
Symptom onset	Symptoms are typically noted in infancy, although milder cases present in childhood. Neonatal hyperammonemia may occur, but is relatively rare.
Symptoms	Spastic diplegia or tetraplegia, opisthotonos, seizures, acquired microcephaly, hepatomegaly, vomiting, anorexia and irritability. Coma and death due to hyperammonemia have been reported.
Natural history without treatment	Developmental delay due to hyperargininemia or hyperammonemia. Neurologic damage can include spasticity, hyperactivity, ataxia, seizures and cerebral atrophy.
Natural history with treatment	Normal outcome may be possible in mild cases, but this has not been proven. Treatment can decrease some of the neurologic symptoms but complete reversal is not likely.
Treatment	Dietary restriction of arginine, protein restriction and supplementation of amino acids other than arginine. Sodium benzoate and/or phenylbutyrate therapy.
Other	Spastic diplegia or tetraplegia.
Physical phenotype	No physical dysmorphism but may be thought to have cerebral palsy.
Inheritance	Autosomal recessive

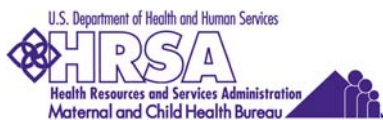
General population incidence	Rare (1: 300,000)
Ethnic differences	No
Population	N/A
Ethnic incidence	N/A
Enzyme location	Liver and erythrocytes
Enzyme function	Terminal enzyme of the urea cycle which hydrolyzes arginine to urea and ornithine to dispose of excess nitrogen.
Missing enzyme	Arginase
Metabolite changes	Increased arginine in plasma, urine and cerebrospinal fluid, increased amino acids (lysine, cystine, ornithine, glutamine) in blood and urine, Hyperammonemia.
Gene	ARG1
Gene location	6p23
DNA testing available	On a research basis only
DNA testing detail	No common mutations known
Prenatal testing	Difficult due to lack of enzyme expression in fibroblasts. Direct fetal blood sampling is a possibility. Prenatal testing by direct DNA testing may be possible if mutations identified. DNA testing may be possible by RFLPs.
MS/MS profile	Arginine and citrulline elevated. Arg – 152-1756 μ M.
OMIM link	www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=207800
Genetests link	www.genetests.org/servlet/access?prg=j&db=genetests&site=&id=12600&fcn=c&qry=22236&res=nous&res=nointl&key=LQ4VNeLSRqKMu&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

Document Info

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