

# Health Care Provider Fact Sheet

<b>Disease Name</b>	<b>Hypermethioninemia</b>
<b>Acronym</b>	MET
<b>Disease Classification</b>	Amino Acid Disorder
<b>Symptom onset</b> <b>Symptoms</b>	Usually asymptomatic in the neonate Sweat, urine, and breath may smell like boiled cabbage; delays in motor skills; intellectual disabilities; muscle weakness; sluggishness; and liver problems can occur.
<b>Natural History w/o treatment</b>	Ranges from asymptomatic to neurologic defects, skeletal deformities, and facial and visual abnormalities. MAT I/III may be benign.
<b>Natural history with treatment</b>	With treatment, many children experience normal growth and learning abilities.
<b>Treatment</b>	The neonate may be prescribed a special formula and later may be restricted to a diet low in protein. Because the diagnosis and management of MET is complex, the primary care physician is advised to manage the baby in close collaboration with a consulting pediatric metabolic disease specialist.
<b>Inheritance</b> <b>General population incidence</b> <b>Ethnic differences</b>	Autosomal recessive RARE, the number of people affected is unknown NA
<b>Missing Enzyme</b>	Methionine adenosyltransferase, glycine N-methyltransferase, <u>or</u> S-adenosylhomocysteine hydrolase
<b>Enzyme location</b>	MAT1A gene = Methionine adenosyltransferase, GNMT gene = glycine N-methyltransferase, AHCY gene = S-adenosylhomocysteine hydrolase
<b>Enzyme function</b>	Each enzyme is a part of a process to convert methionine into a usable amino acid. Methionine adenosyltransferase converts methionine into S-adenosylmethionine (AdoMet); glycine N-methyltransferase begins the next step in the process by converting AdoMet into S-adenosyl homocysteine (AdoHcy); S-adenosylhomocysteine hydrolase converts AdoHcy into homocysteine which is then converted into the amino acid, cysteine
<b>MS/MS Profile</b>	Methionine – elevated
<b>OMIM Link</b> <b>Genetests Link</b> <b>Support Group</b>	<a href="http://www.omim.org">www.omim.org</a> ID *610550 <a href="http://www.genetest.org">www.genetest.org</a> Genetics Home Reference <a href="http://www.ghr.nlm.nih.gov/ghr/page/home">www.ghr.nlm.nih.gov/ghr/page/home</a> Save Babies through Screening Foundation <a href="http://www.savebabies.org">www.savebabies.org</a> Children Living with Inherited metabolic Diseases (CLIMB) <a href="http://www.climb.org.uk/">www.climb.org.uk/</a>