

Fatty Acid Metabolism Disorders

Medium Chain Acyl co-A Dehydrogenase Deficiency (MCAD)

Medium chain acyl co-A dehydrogenase deficiency (MCAD) is an inborn error of fatty acid oxidation that can cause significant morbidity and mortality in the newborn. It usually presents in infancy or early childhood with hypoketotic hypoglycemia and encephalopathy after an intercurrent illness and/or period of poor oral intake. Approximately 20% of infants with MCAD die before diagnosis, and a substantial proportion of the survivors have significant residual problems from an initial crisis. Children who survive the initial crises may have developmental delay, seizures, speech/language delays, chronic muscle weakness, failure to thrive, cerebral palsy and attention deficit disorder.

Inheritance: Autosomal recessive

Estimated Incidence: 1:16,000

Abnormal Screen Result: Primary Markers
Elevated C8 (octanoyl carnitine)
Elevated C8/C10

Secondary Markers
Elevated C6 (hexanoyl carnitine)
Elevated C10 (decanoyl carnitine)
Elevated C10:1 (decenoyl carnitine)

Method of Notification: All abnormal results where the C8 is elevated are called to physician of record.

If the C8/C10 ratio is elevated, but the C8 is within normal limits, the physician of record is notified by mail.

Isolated elevations of secondary markers have no clinical significance and are not reported.

Next Steps if Abnormal: See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.

Neonatal Presentation: Usually none

Treatment: Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop.

Infants with MCAD must be fed at least every four hours, including at night. **Infants with MCAD should not be fed formulas that have**

medium chain triglycerides (MCT) as a fat source if a safe alternative is available. (A list of formulas that contain MCT as a fat source is included on this page.) Feeding intervals can be lengthened as the infant gets older.

Carnitine supplementation if helpful.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with MCAD. These outcomes include hypoketotic hypoglycemia, vomiting, lethargy, seizures and coma. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent hypoglycemic episodes. Blood glucose may be monitored as a precaution.

Premature/sick infants—Some special formulas and breast milk fortifiers fed to premature/sick infants contain medium chain triglycerides (MCT) as the primary fat source. These feedings may cause false elevations of some acyl carnitines analyzed in MCAD screening, particularly C8, C10:1 and C8/C10.

MCT Containing Formulas

Formula Type	Product Name--MCT is Primary Fat Source
Formulas for Preterm Infants	Enfamil Premature Lipil Similac Special Care 20, Similac Special Care 24, Similac Special Care 30
Breast Milk Fortifiers	Enfamil Human Milk Fortifier Similac Human Milk Fortifier
Free Amino Acid Formulas	Vivonex Pediatric
Protein Hydrolysate Formulas	Pregestimil, Pregestimil Lipil Peptamen Junior, Peptamen Junior Fiber, Peptamen Junior Prebio Pediatric Peptinex DT, Pediatric Peptinex DT with Fiber
Formulas for Children or Special Circumstances	Portagen Monogen
Modular Components	MCT Oil Product 3232A

This list is not all inclusive. Presence on this list does not constitute any endorsement by DHEC.

Medium Chain Ketoacyl co-A Thiolase Deficiency

Medium chain ketoacyl co-A thiolase deficiency (MCKAT) is an inborn error of fatty acid oxidation. One infant with this disorder has been detected worldwide. This male neonate presented with vomiting, dehydration, metabolic acidosis, liver dysfunction, and terminal rhabdomyolysis with myoglobinuria.

Inheritance:	Unknown
Estimated Incidence:	Extremely rare
Abnormal Screen Result:	Elevated C8 (octanoyl carnitine) Elevated C6DC (adipyl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Vomiting, dehydration, metabolic acidosis, liver dysfunction
Treatment:	Only known affected infant died at 13 days of life. Presumed treatment is same as that for other fatty acid metabolism disorders: Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Short Chain Acyl co-A Dehydrogenase Deficiency (SCAD)

Short chain acyl co-A dehydrogenase deficiency (SCAD) is an inborn error of fatty acid oxidation. Outcome in affected persons have been quite variable. Infants may have seizures, poor feeding, progressive muscle weakness, developmental delay and hypotonia.

Inheritance:	Autosomal recessive
Estimated Incidence:	1:40,000 to 1:100,000
Abnormal Screen Result:	Elevated C4 (butyryl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Poor feeding, vomiting, lethargy, seizures, hypotonia, hepatomegaly
Treatment:	<p>Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop.</p> <p>Feed at least every four hours, including at night. Feeding intervals can be lengthened as the infant gets older.</p> <p>Carnitine supplementation if low. Riboflavin trial.</p>

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Medium and Short Chain Acyl co-A Dehydrogenase Deficiency (M/SCHAD)

Medium and short chain acyl co-A dehydrogenase deficiency (M/SCHAD) is an inborn error of fatty acid oxidation caused by deficiency of 3-OH acyl co-A dehydrogenase. Therefore, it is also referred to as 3-OH acyl co-A dehydrogenase deficiency (HADH).

Inheritance:	Autosomal recessive
Estimated Incidence:	Unknown, thought to be rare
Abnormal Screen Result:	Elevated C4OH (3-OH butyryl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Poor feeding, vomiting, lethargy, hypoglycemia. Plasma insulin may be elevated.
Treatment:	<p>Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop.</p> <p>Feed at least every four hours, including at night. Once child is older, may need cornstarch supplementation at bedtime to maintain blood glucose levels overnight.</p> <p>Carnitine supplementation if helpful. Consider medication for infants with documented hyperinsulinism.</p>

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Dienoyl co-A Reductase Deficiency

Dienoyl co-A reductase deficiency is a very rare defect in fatty acid oxidation. One African-American infant with this disorder has been described. Infant had hypotonia, small ventricular septal defect, short extremities and microcephaly; however, the relationship of phenotype to this disorder is not known.

Inheritance:	Presumed autosomal recessive
Estimated Incidence:	Unknown, thought to be very rare
Abnormal Screen Result:	Elevated C10:2 (decadienoyl carnitine)
Method of Notification:	All results called to the physician of record
Next Steps If Abnormal:	Potential medical emergency. See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies could include plasma total and free carnitines, plasma acylcarnitines and urine organic acids. In addition, repeat acyl carnitine profile on filter paper and send to the DHEC laboratory.
Neonatal Presentation:	Possible hypotonia, small ventricular septal defect, short extremities and microcephaly
Treatment:	Proposed treatment includes avoidance of fasting. Supplementation with high energy drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Other suggested treatment includes reduction of long chain fat in the diet and supplementation with MCT and carnitine.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Long Chain 3-OH Acyl co-A Dehydrogenase Deficiency (LCHAD) Trifunctional Protein Deficiency (TFP)

Long chain 3-OH acyl co-A dehydrogenase is one of three enzymes in trifunctional protein deficiency (TFP). Deficiencies of these enzymes overlap clinically. Affected persons may present with hypoketotic hypoglycemia, hepatic encephalopathy and muscle weakness usually associated with cardiomyopathy. Other features include rhabdomyolysis, myoglobinuria and peripheral neuropathy. They may also show retinal pigmentation with vision loss in childhood. Symptoms may present as early as the first days of life.

Inheritance:	Autosomal recessive
Estimated Incidence:	Unknown
Abnormal Screen Result:	Primary Marker Elevated C16-OH (3-OH palmitoyl carnitine) Secondary Markers Elevated C14:1 (tetradecenoyl carnitine) Elevated C16 (palmitoyl carnitine) Elevated C18:1 (oleyl carnitine) Elevated C18:1-OH (3-OH oleyl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Hypoketotic hypoglycemia. Some infants will have hepatic encephalopathy and muscle weakness associated with cardiomyopathy.
Treatment:	Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night. May need cornstarch supplementation via tube feeding overnight in older infancy/childhood. Fat restricted diet with use of MCT (medium chain triglyceride) oil as fat source. Essential fatty acid supplementation. DHA (docosahexanoic acid) supplementation to prevent retinal degeneration may be used. Carnitine supplementation if helpful.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

HELLP Syndrome/AFLP—HELLP syndrome (hemolysis, elevated liver enzymes, low platelets)/AFLP (acute fatty liver of pregnancy) occurs in 20% of pregnancies where the fetus is affected by LCHAD. TFP is thought to be a less common cause of HELLP syndrome.

Very Long Chain Acyl co-A Dehydrogenase Deficiency (VLCAD)

Very long chain acyl co-A dehydrogenase deficiency (VLCAD) is an inborn error of fatty acid oxidation. Infants may have hypoketotic hypoglycemia, hypotonia, hepatic dysfunction and cardiomyopathy. 20% of affected persons present as adolescents or adults with muscle fatigue, rhabdomyolysis and myoglobinuria triggered by exercise or fasting.

Inheritance:	Autosomal recessive
Estimated Incidence:	Unknown
Abnormal Screen Result:	Primary Markers Elevated C14:1 (tetradecenoyl carnitine) Elevated C14:1/C12:1 Elevated C14:1/C16 Secondary Markers Elevated C14 (tetradecanoyl carnitine) Elevated C14:2 (tetradecadienoyl carnitine) Elevated C16 (palmitoyl carnitine) Elevated C18:1 (oleyl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Hypoketotic hypoglycemia, hepatic dysfunction, hypotonia and cardiomyopathy
Treatment:	Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night. May need cornstarch supplementation via tube feeding overnight in older infancy/childhood. Fat restricted diet with use of MCT (medium chain triglyceride) oil as fat source. Essential fatty acid supplementation. Carnitine supplementation if helpful.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid

oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Multiple acyl coA Dehydrogenase Deficiency/Glutaric Aciduria Type II (MAD/GA II)

Multiple acyl coA dehydrogenase deficiency (MAD) or glutaric aciduria type II (GA II) is a fatty acid oxidation disorder caused by a deficiency of electron transfer flavoprotein (ETF) or ETF-ubiquinone oxidoreductase. These enzymes transfer electrons from the first step in β -oxidation to the electron transport chain. There are three described types of GA II: neonatal onset with congenital anomalies, neonatal onset without congenital anomalies, and mild/late onset. Outcome for infants with GA II and congenital anomalies is extremely grave.

Inheritance: Autosomal recessive

Estimated Incidence: Unknown

Abnormal Screen Result: Primary Markers
Elevated C4 (butyryl carnitine)
Elevated C5 (isovaleryl carnitine)

Secondary Markers
Elevated C6 (hexanoyl carnitine)
Elevated C8 (octanoyl carnitine)
Elevated C10 (decanoyl carnitine)
Elevated C16 (palmitoyl carnitine)
Elevated C18:1 (oleyl carnitine)

Method of Notification: All abnormal results called to physician of record

Next Steps if Abnormal: **Potential medical emergency.** See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.

Neonatal Presentation: Neonatal onset with congenital anomalies—prematurity, hypotonia, metabolic acidosis, cystic kidneys, facial dysmorphisms, rocker bottom feet

Neonatal onset without congenital anomalies—hypotonia, tachypnea, metabolic acidosis, hypoglycemia, sweaty feet odor, cardiomyopathy

Treatment: Neonatal onset with congenital anomalies—no treatment effective

Neonatal onset without congenital anomalies—treatment probably not effective

Mild/late onset—Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop.

Feed at least every four hours, including at night. May need cornstarch supplementation via tube feeding overnight in older infancy/childhood. May be prescribed diet restricted in fat, controlled in protein. Riboflavin supplementation if helpful.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Carnitine Uptake/Transport Deficiency

In carnitine uptake/transport deficiency (CUD), carnitine transport across the plasma membrane is inhibited. The reduction in carnitine limits the formation of acylcarnitine and subsequently limits energy production. Skeletal and heart muscle tissues are particularly affected in this process.

Inheritance:	Autosomal recessive
Estimated Incidence:	1:100,000
Abnormal Screen Result:	Low free carnitine
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	Potential medical emergency. See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Tachycardia, hepatomegaly, reduced muscle tone, poor feeding
Treatment:	Carnitine supplementation. Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night. Feeding regimen may be adjusted when the infant is older. May be prescribed diet restricted in fat.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Maternal CUD—In some newborns, the low free carnitine is reflective of maternal CUD.

Carnitine Palmitoyl Transferase I Deficiency (CPT I)

Carnitine palmitoyl transferase I (CPT I) is necessary for the conversion of long chain fatty acyl co-A molecules to their corresponding acylcarnitine molecules. Deficiency of this enzyme reduces the availability of acylcarnitines for transport into the mitochondrial matrix for fatty acid oxidation.

Inheritance:	Autosomal recessive
Estimated Incidence:	Unknown, thought to be more common in persons of Inuit or Hutterite origins
Abnormal Screen Result:	Elevated Free Carnitine/C16 + C18
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	Potential medical emergency. See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Seizures, hepatomegaly, hypoketotic hypoglycemia
Treatment:	Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night. Feeding regimen may be adjusted when the infant is older. May be prescribed diet restricted in fat. MCT (medium chain triglyceride) oil may be used as a fat source after diagnosis is clearly established.

Carnitine is contraindicated in treatment of CPT I.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

HELLP Syndrome/AFLP—HELLP syndrome (hemolysis, elevated liver enzymes, low platelets)/AFLP (acute fatty liver of pregnancy) can occur in pregnancies where the fetus is affected by CPT I.

Carnitine Palmitoyl Transferase II Deficiency (CPT II)

Carnitine palmitoyl transferase II deficiency (CPT II) is a disorder of fatty acid transport. In classic CPT II, the onset is during adolescence or early adulthood. It presents with muscle weakness, pain and myoglobinuria usually prompted by exercise, but sometimes by fasting, infection or stress. Renal failure from myoglobinuria occurs in 25% of affected persons. The neonatal type, hepatocardiomyopathy CPT II, is extremely rare. Symptoms include hypoketotic hypoglycemia, hepatomegaly, skeletal muscle involvement and marked lipid accumulation in muscle. These infants may also have dysmorphic features.

Inheritance:	Autosomal recessive
Estimated Incidence:	Unknown
Abnormal Screen Result:	Elevated C16 (palmitoyl carnitine) Elevated C18:1 (oleyl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	Potential medical emergency. See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Classic CPT II—none Hepatocardiomyopathy CPT II—hypoketotic hypoglycemia, hepatomegaly, skeletal muscle involvement, marked lipid accumulation in muscle, dysmorphic features.
Treatment:	Classic CPT II—Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night. Feeding regimen may be adjusted when the infant is older. May be prescribed diet restricted in fat. MCT (medium chain triglyceride) oil may be used as a fat source after diagnosis is clearly established. Hepatocardiomyopathy CPT II—treatment probably not effective

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.

Carnitine/Acylcarnitine Translocase Deficiency (CACT)

Carnitine/acylcarnitine translocase deficiency (CACT) is a disorder of fatty acid and carnitine transport. Two types have been described, one with neonatal onset and the other with onset later in infancy/early childhood. In neonatal onset CACT, the affected infant presents with a metabolic crisis that often results in death from cardiopulmonary complications and/or liver failure. When the onset is later in infancy/early childhood, the affected person presents with hypoglycemia, but not cardiomyopathy.

Inheritance:	Autosomal recessive
Estimated Incidence:	Unknown, thought to be very rare
Abnormal Screen Result:	Elevated C16 (palmitoyl carnitine) Elevated C18:1 (oleyl carnitine)
Method of Notification:	All abnormal results called to physician of record
Next Steps if Abnormal:	Potential medical emergency. See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist for further instructions. Repeat acyl carnitine profile as soon as possible on filter paper. Initiate treatment and diagnostic evaluation as recommended by specialist.
Neonatal Presentation:	Neonatal onset CACT—hypoketotic hypoglycemia, hyperammonemia, hypotonia, liver dysfunction, cardiomyopathy Later infancy/early childhood onset CACT—none
Treatment:	Neonatal onset CACT—treatment probably not effective Later infancy/early childhood onset CACT—Avoid fasting. Supplementation with high energy carbohydrate drinks during periods of illness. Seek medical attention if these liquids are refused. Oral rehydration fluids are not adequate. If oral feedings are not possible, then intravenous fluids with concentrated dextrose should be started without waiting for symptoms of decompensation to develop. Feed at least every four hours, including at night. Feeding regimen may be adjusted when the infant is older. May be prescribed diet restricted in fat. MCT (medium chain triglyceride) oil may be used as a fat source after diagnosis is clearly established.

Special Considerations

Fasting/infection/intercurrent illness—Parents must clearly understand that prolonged fasting and infection/intercurrent illnesses can cause serious complications in an infant with a fatty acid oxidation disorder. Infants experiencing stressful events such as trauma, infections/illnesses, and even receiving immunizations should be monitored to prevent metabolic decompensation.