



## HELLP, AFLP, and FAODs

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**HELLP** is an acronym for **h**emolysis (destruction of red blood cells), **e**levated **l**iver **e**nzymes and **l**ow **p**latelets, and **AFLP** is an acronym for **a**cute **f**atty **l**iver of **p**regnancy. These pregnancy complications can develop in the third trimester of pregnancy, and the cause is usually unknown. HELLP syndrome is considered a complication of severe pre-eclampsia and is estimated to occur in 0.1-0.6% of all pregnancies and in 4-12% of women with a diagnosis of pre-eclampsia. Recurrence risk for HELLP syndrome in later pregnancies is 3 to 27%.

AFLP is a more severe disorder with an increased risk for maternal death. It occurs in approximately 1 in 13,000 pregnancies. Women with AFLP initially may have abdominal pain, nausea and vomiting and can rapidly develop liver failure, bleeding disorders and encephalopathy (a brain disorder). There is limited information regarding the risk of recurrence of this disorder.

Both HELLP syndrome and AFLP are treated by delivery of the pregnancy, after which the symptoms usually improve. There is overlap between the symptoms and lab findings of HELLP syndrome and AFLP so that it can sometimes be difficult to distinguish which disorder an individual has. Some experts believe that preeclampsia, HELLP syndrome and AFLP represent different stages of the same disease and may share some common causes.

### What are FAODs?

**FAOD** is an acronym for **f**atty **a**cid **o**xidation **d**isorder. FAODs are genetic disorders that result from the inability to breakdown fats in the body. After a meal, the body uses the energy from the food directly, and stores what is unused as glycogen in the liver. If the short-term energy supply of the body is depleted (such as fasting overnight), fatty acids are broken down to create energy. Skeletal muscle and the heart use fatty acids as a major source of energy. There are many different enzymes involved in the breakdown of fats for energy. If any of the enzymes is not working properly, the result is an FAOD. Each enzyme is responsible for a different FAOD. The most common FAOD is called MCAD (**m**edium **c**hain **a**cyl-**C**oA **d**ehydrogenase) deficiency.

Individuals with FAODs can come to medical attention because of low blood sugar, lethargy, sudden infant death, and Reye-like syndrome. Individuals with FAODs can also have hypotonia (poor muscle tone) and cardiomyopathy (heart function defect). Severe complications are more likely to occur after an illness or prolonged period of fasting. Sometimes individuals with FAODs have no symptoms that bring them to medical attention. Currently, most states in the US provide mandated newborn screening for FAODs at birth in order to identify children with these disorders and prevent long-term complications.

FAODs are inherited in an autosomal recessive pattern in families. In autosomal recessive inheritance, an affected individual has two copies of a gene that are not working properly. Therefore, the parents of affected individuals are most likely carriers (occasionally the parent of an affected individual may also have the same FAOD). Individuals who are carriers for FAODs have one copy of the gene working properly and one not working properly. Male carriers of FAODs show no symptoms, and female carriers are at risk for pregnancy complications (see below). If two individuals are carriers for the same FAOD there is a 1 in 4 (or 25%) chance with each pregnancy to have a child with a FAOD. Prenatal testing by chorionic villus sampling (performed at 10 to 12 weeks in the pregnancy) or amniocentesis (performed at 15 weeks or greater in the pregnancy) is available for many families. Your doctor or genetic counselor can discuss these options with you in more detail.

**Are FAODs treatable?**

Yes. Individuals with FAODs should avoid fasting. This means newborns should not go more than 2 to 3 hours without food. Older children and adults may go longer before they reach a fasting state. Illnesses, such as the flu, may necessitate medical treatment or hospitalization. In addition, changes in the diet or dietary supplements may be required to reduce the need of the body to break down the fats. Consultation with a dietician and a medical geneticist familiar with treatment of FAODs is important for affected individuals, as other therapies may be recommended based on the findings in a specific case.

**How are HELLP syndrome, AFLP and FAODs related?**

Women who are carriers for a FAOD, specifically a condition called LCHAD (long chain 3-hydroxyacyl coenzyme A dehydrogenase) deficiency, are at risk for pregnancy complications such as HELLP syndrome and AFLP if the unborn child is affected with LCHAD deficiency. A recent study performed by researchers at Harvard University revealed that maternal liver disease, namely AFLP, was found in 16% of pregnancies when the baby had LCHAD deficiency, compared to less than 1% of pregnancies when the baby did not have LCHAD deficiency. Isolated pre-eclampsia was not associated with an increased risk for a FAOD in the baby. The risk for a FAOD in the baby when a woman has HELLP or AFLP is not yet clear; however earlier studies have revealed that if a woman has HELLP syndrome during pregnancy, there is a 2% risk of LCHAD deficiency in the baby. In women with AFLP during pregnancy, the risk of a child affected with LCHAD deficiency is 15-20%. It is likely that when considering all FAODs, the risks would be even higher.

**What can I do if I have HELLP syndrome or AFLP in my pregnancy?**

Women with HELLP syndrome and AFLP should consider testing their offspring for the presence of a FAOD. In addition, since affected individuals may never show symptoms, parents of a child affected with a FAOD may consider diagnostic testing for FAODs. Many states offer expanded newborn screening which includes several FAODs, so women with HELLP syndrome and AFLP may want to check with their OB/GYN regarding what testing is available.