

Commonly asked questions

1. What is IVA?

IVA is also known as isovaleric acidemia and is an organic acid disorder caused by a defect in the metabolism of a specific essential amino acid, leucine. The inability to completely metabolize this amino acid leads to a build up of toxic intermediate chemicals. This is often exacerbated when the body is stressed (e.g. fasting, operations or infections). During these times the body breaks down its own proteins to supply needed energy and as a result, the amino acids are metabolized into the toxic intermediates.

2. How and when will we know if my baby has IVA?

If your baby's newborn screening result showed a markedly elevated C5 levels, he or she probably has IVA. The newborn screening test will be repeated and additional tests will be undertaken to help determine whether or not your baby has IVA. Typically, the results of these tests take up to 4 days to come back. Depending on the test results, additional testing can take a variable amount of time to confirm the diagnosis. In a very small minority of cases it can be difficult to determine whether or not a child is affected.

3. How did my baby get this?

IVA is an autosomal recessive genetic disorder. This means that your baby has two abnormal mutated genes, one from the mother and one from the father. Having only one mutated gene (a carrier) does not affect a person at all.

4. What does it mean for my child?

If your baby has IVA, he or she will have to have a special protein restricted diet. Most children with this condition also take glycine and carnitine, a mild supplemental medicine. If your child becomes ill, it may well be necessary early in the illness (i.e. when it might be considered mild), to further restrict the protein intake for a short period of time or even to provide extra energy in the form of glucose through addition to food or, if necessary, by intravenous infusion. By treating your baby this way it is possible to generally prevent the worst effects of these conditions. However, babies and children with IVA are at risk from serious effects such as mental retardation, loss of control of movement or even death if allowed to get sick throughout childhood. Therefore, it is important to maintain vigilance, consider every illness seriously and hospitalize for specialized treatment early. Some children, despite the best treatment and care possible, will still have some delay though this will be significantly less than if your child is not treated as described above.

5. What is the treatment? Does it work? Is the diet difficult to do/expensive?

IVA is primarily treated by a protein-restricted diet and special formula composed of amino acids. The special formula, which will keep your child well, is typically ordered through your metabolic clinic where the metabolic nutritionist will ensure that you are confident in preparing it. The formula can be expensive; however, your metabolic clinic will assist you in obtaining it through your health care provider or state agency.

6. What about my other children/future children?

As IVA is an inherited condition it is essential to have your other children tested. Children from the same father and mother as the affected infant have a 1 in 4 (25%) chance of having the same condition. Your other children can appear healthy and still have the disorder. If they have IVA, successfully having weathered illnesses in the past is no guarantee that an illness in the future will not have serious consequences.

Since there is a risk for having a future child with IVA it is important to let your obstetrician and pediatrician know that you have a child with IVA if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.