

Normal repeat newborn screening result.

If the first screen showed an elevated level of leucine, a normal second screen result is reassuring BUT does not rule out MSUD. Therefore, the metabolic physician may want to continue treating the baby as though he/she has MSUD while awaiting the results of more definitive tests.

Once the metabolic team has confirmed that the infant does not have MSUD, it is essential to reassure the family that their baby is well and that they should treat their baby as entirely normal. Many people can be traumatized by a false positive result and counseling may be appropriate. If the metabolic physician remains concerned, however, then he/she will discuss this further with you and may decide to continue with early intervention if the baby becomes sick. It is important to remember that this does not mean that the baby has MSUD but only that the metabolic doctor is taking a cautious approach until definitive results are available to keep the baby safe and well.

Abnormal repeat newborn screen result.

An elevated leucine on the second sample is very suspicious of MSUD and further evaluation by the metabolic doctor is definitely required. The baby must be treated as though he/she has MSUD while definitive testing is carried out.

See **leucine markedly elevated, probable MSUD** discussions in first newborn screening result section.