



Online learning factsheet: isovaleric acidaemia (IVA)

	Isovaleric acidaemia (IVA)
Problem breaking down	Leucine amino acid
Incidence	1:155,000 (EU data)
Early effects	<p>Early signs may include:</p> <ul style="list-style-type: none"> • vomiting • excessive sleepiness • floppiness • rapid breathing
Untreated	<ul style="list-style-type: none"> • coma and permanent brain damage or death in some cases • some babies with IVA have problems within a few days of birth; others become unwell at a few months or years of age, perhaps during a minor illness
Treatment	<ul style="list-style-type: none"> • a special low protein diet, and carnitine and glycine • a different regime is required when the child is ill, and they may need to be hospitalised
Notes	<ul style="list-style-type: none"> • babies can become suddenly and seriously ill and will need to be seen immediately – they may be offered transfer via ambulance • babies cannot be exclusively breastfed
Online information	<p>http://newbornbloodspot.screening.nhs.uk/iva</p> <p>http://www.expandedscreening.org/site/home/metabolic-iva-introduction.asp</p>



KEY MESSAGES

- Screening for the four additional disorders will be rolled out across England from January 2015
- They are all inherited metabolic diseases (IMDs) like PKU and MCADD
- They are very rare disorders (much rarer than the disorders currently screened for)
- No additional blood spots are required (but four good quality spots are needed)
- No specific repeat tests are needed
- Some babies with MSUD and IVA can become suddenly and seriously ill and will need to be seen immediately
- The six IMDs (PKU, MCADD, MSUD, IVA, GA1 and HCU) can only be declined as a group and not individually
- Like the existing disorders, parents can expect to receive normal results within 6-8 weeks
- There are no particular ethnicities associated with the additional disorders
- Parent leaflets, videos and further information is available at <http://newbornbloodspot.screening.nhs.uk/imd>
- Information available in Screening Tests for You and Your Baby at <http://newbornbloodspot.screening.nhs.uk/leaflets>