Maple Syrup Urine Disease
What is Maple Syrup Urine Disease (MSUD)?

- genetic disease that causes problems in protein breakdown
- the urine and earwax of people with MSUD have the characteristic odour of maple syrup
What is missing in MSUD?

- the enzymes that normally break down Leucine, Isoleucine & Valine (The branched chain amino acids)
What does the enzyme problem cause?

• BCAAs pile up
• ketoacids form
• ketoacids in the blood are toxic to the brain
How do people get MSUD?

- One MSUD gene is inherited from both parents by recessive inheritance

- Baby therefore has to get a ‘double dose’ of the gene to be affected with MSUD
How common is MSUD?

- MSUD occurs in about 1 per 180,000 newborns in the US
- may be as frequent as 1 per 176 newborns in selected populations such as the Mennonites of Pennsylvania
- more prevalent in populations with a high frequency of intermarriage
How are people with MSUD diagnosed?

• Symptoms such as poor feeding, poor weight gain and sluggishness develop in newborns aged 4-7 days

• neurological problems such as seizures and brain swelling develop rapidly

• a blood test confirms MSUD

• some States test all babies at birth for MSUD
Effects of untreated classic MSUD

- Mental retardation
- Walking and speech problems
- Seizures
- Death
What do ‘high levels’ look like?

- irritability
- confusion
- sleepiness
- staggering

- hallucinations
- slurred speech
- vomiting
- unusual breathing
What causes high levels?

- Colds, flu, infections, surgery and stress
- Any signs of illness require initiation of the sick day plan
What do high levels cause?

- Left untreated, high levels can cause the person with MSUD to lose consciousness, go into a coma and die
What can be done?

- The damage from high levels of BCAAs in infancy is irreversible.
- Further damage can be prevented by strict dietary treatment.