NEWBORN SCREENING- Dr Assim Javaid and Dr Graham Shortland

2:15 The heel prick test

- Sickle cell disease to prevent the complications
- Cystic fibrosis
- Congential hypothyroidism

Picks up deficit in thyroxine
Test for TSH
Could be some congenital absence of the thyroid which could be missed

PKU
9:00 MCADD Medium chain acyl-CoA dehydrogenase deficiency

10:51
glutaric aciduria type 1 (GA1)
isovaleric acidaemia (IVA)

homocystinuria (pyridoxine unresponsive) (HCU)

11:50 it is important that the baby picked up is seen quickly
Can present with coma, apnoea,
Speak urgents
12:50 MSUD maple syrup urine disease
Management can prevent later decompensation

13:40 Babies may not have been screened, may have moved into the area.
Screening except cystic fibrosis can be done up to 12 months

15:30 Keep a look out for changes in the screening programme
Newborn screening NHS website
https://www.nhs.uk/conditions/pregnancy-and-baby/newborn-blood-spot-test/

17:20 Key points

- If you have a clinical suspicion do the tests to confirm
- May not have had screening and also may be false negative
- Keep on top of changes
- If you have any concerns about metabolic disease speak to a senior and your nearest metabolic disease centre

21:05 Follow Wilson criteria
https://www.ldh.nhs.uk/wp-content/uploads/2018/04/criteria_for_screening.pdf