

A better test for galactosaemia

Innovation and Service Improvement
MAHSE Research Day 9th June 2014

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Content

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 - What is galactosaemia?
 - Why is it important to diagnose it very quickly?
- Current galactosaemia test
- What do we want from a new test?
- The new test
- Benefits of the new test
- Conclusions

What is galactosaemia?

Healthy people



Diet Milk feeds (**Galactose**)

Galactose

Galactose 1-phosphate

*Galactose 1-phosphate
uridylyltransferase*

Glucose 1-phosphate

What is galactosaemia?

Galactosaemia



Diet Milk feeds (**Galactose**)

Galactose

Galactose 1-phosphate

*Galactose 1-phosphate
uridylyltransferase*



Glucose 1-phosphate

The reasons galactosaemia is hard to diagnose

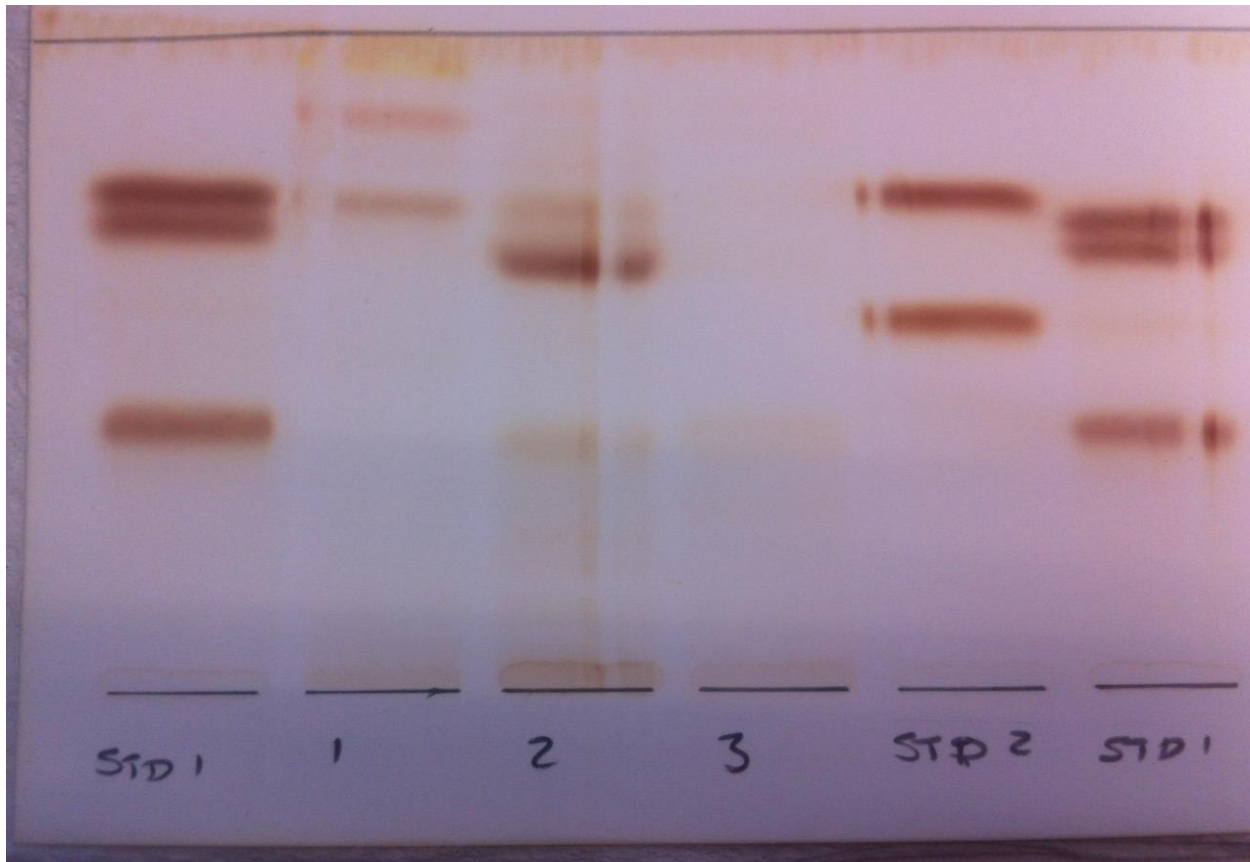
- Symptoms include:
 - Sickness and diarrhoea
 - Liver failure
 - Septicaemia (severe bacterial infection)
 - Delayed growth
- Reasons why galactosaemia is hard to diagnose:
 - The symptoms are similar to other conditions
 - The rarity of the condition puts it low on a doctor's radar

Left untreated galactosaemia
can be fatal

Current way of testing for galactosaemia: urinary reducing substances

- Measures sugars in urine
- Time-consuming and manual
- Results take three weeks
- Prone to false positive results in liver failure
- Testing involves visual inspection of results

Urinary reducing substances example

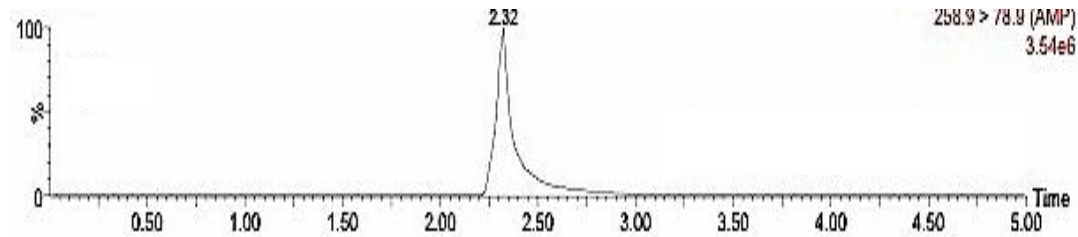


Requirements for the ideal new fast test

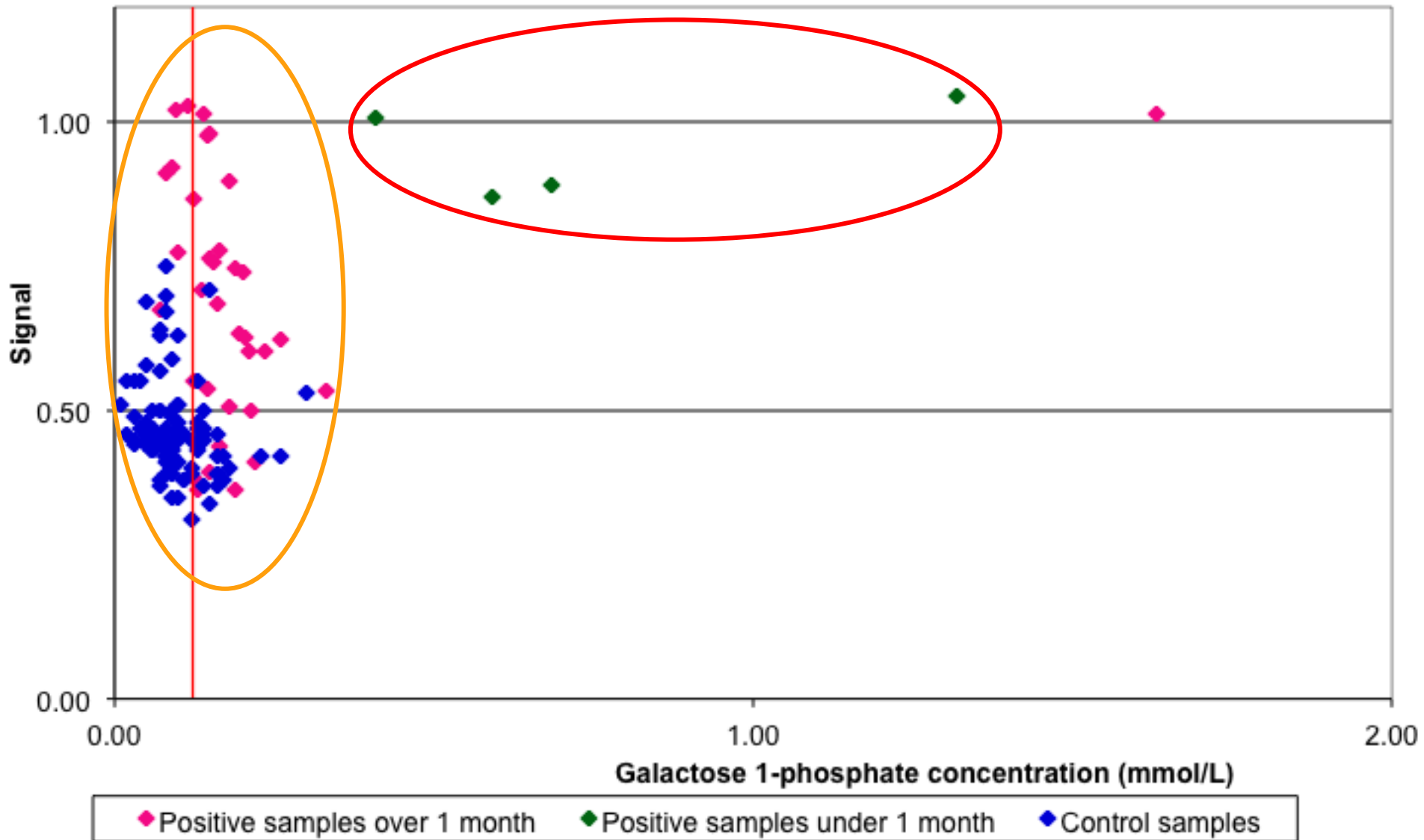
- Highly sensitive and highly specific
- Uses dried blood spot samples
- Same day result
- Cost effective (cheaper than previous method)
- Suitable for the testing of individual samples
- Easy sample preparation



The new fast test



Patient data



Benefits of the new fast test

- Not prone to false positive results in liver failure
- Easier to collect heel prick samples than blood samples from a vein
- Easy sample preparation
- More accurate and sensitive results
- Same day results



Conclusions

- Fast diagnosis of galactosaemia is vital
- A new fast test using dried blood spots for galactosaemia has been developed
- The new test is not prone to false positives in liver failure
- The new test improves galactosaemia diagnosis for very unwell babies by providing same day results

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