BIMDG
British Inherited Metabolic Disease Group

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GLYCOGEN STORAGE DISEASE (GSD) TYPE I
MANAGEMENT OF NEWBORN BABIES WITH A FAMILY HISTORY

- **Please read this carefully as there is a risk of death and serious complications in the newborn period.**

BACKGROUND

Glycogen storage disease type I is an inherited disorder that prevents the breakdown of glycogen stored in the liver; it is caused by glucose-6-phosphatase deficiency. Patients have a severe tendency to hypoglycaemia. This generally occurs after fasting for about 2 hours, accompanied by raised lactate concentrations. Hepatomegaly and raised triglyceride concentrations are expected but may be less obvious than in older patients.

GSD I is an autosomal recessive disorder, so the recurrence risk is 1:4 if a couple have previously had an affected child. Surprisingly, many patients only have mild symptoms in the neonatal period but they do become hypoglycaemic, so it is desirable to establish whether they are affected as soon as possible.

The diagnosis is best confirmed by mutation analysis & it may be possible for this to be done within 2 weeks if the mutation(s) are known in the index case & the case is discussed in advance with the molecular genetic laboratory. However, it should be possible to get an earlier answer by monitoring the blood glucose concentration.

MANAGEMENT

1. In advance, discuss plans with the parents & the molecular genetics laboratory.
2. Within 10-15 minutes of birth, start feeding the baby with standard infant formula at **80ml/kg/day every 2 hours**. If this is not taken, pass a nasogastric tube & give through this. Subsequently, the baby can be put to the breast if so wished.
3. Admit the baby to the neonatal unit, weigh and examine for liver size.
4. Inform the consultant on call at the Regional Metabolic Centre about the baby.
5. The baby should be fed no less frequently than every TWO HOURS at 80ml/kg/day of standard infant formula. A nasogastric tube may well be required. If breastfeeding give the volume of formula followed by breast feeding ad lib.
6. Monitor pre-feed blood glucose levels and confirm with blood glucose levels measured in the laboratory (bedside point-of-care testing machines can be unreliable at low levels). Aim to keep the blood glucose >3.5 mmol/L & certainly >3.0 mmol/L.

If the blood glucose is consistently low on two hourly feeding, discuss with the regional metabolic centre and try feeding at 80ml/kg/day given as 16 feeds a day every 90 minutes. If this is required it is highly likely the baby is affected.

7. Measure plasma lactate, triglycerides, cholesterol and uric acid, which are often increased in children with GSD I. Send blood for mutation analysis, though glucose monitoring will probably indicate if the baby is affected before genetic results are back. Consider inserting an intravenous cannula, in case needed.

8. If 2 hourly feeding is tolerated without hypoglycaemia then, after 24 hours on Day 2, the baby can be fed at 120ml/kg/day with the feeding interval being increased in 30 minute increments, with pre-feed blood glucose monitoring.

    - For 2½ hourly feeding give 12.5ml/kg/feed
    - For 3 hourly feeding give 15ml/kg/feed
    - For 3½ hourly feeding give 17.5mlskg/feed
    - For 4 hourly feeding give 20ml/kg/feed

    If the baby is normoglycaemic 3-4 hours after feeding then he/she is unlikely to be affected by GSD I and the baby can be fed normally.

    If the baby is not tolerating feeds well on day 2 then stretching the feed interval can be delayed until day 3 when the baby's feeding volume could be increased to 150ml/kg/day.

9. If affected, the fasting tolerance may be between 1.5-2 hours. Further management advice should be obtained from the regional metabolic consultant and specialist metabolic dietitians.

PROBLEMS

10. If the baby does not tolerate feeding, intravenous glucose will be needed to prevent hypoglycaemia until feeding can be established. **Give 10% glucose at 5ml/kg/hr.** KCl can be added if appropriate & 10% glucose 0.45% NaCl can be substituted based on plasma electrolytes & the policies of the neonatal unit.

11. If there is **asymptomatic hypoglycaemia**, give the baby a feed, if necessary through a nasogastric tube. If there is **symptomatic hypoglycaemia**, give an intravenous bolus of 5ml/kg 10% glucose followed by an infusion at 5ml/kg/hr. If there is no intravenous access, give glucogel into the buccal cavity whilst siting a cannula.