APPENDIX R
SCREENING FOR GENETIC METABOLIC DISORDERS

Extract from: Christodoulou J, Wilcken B. Perimortem laboratory investigation of genetic metabolic disorders. Seminars in Neonatology 2004;9(4):275-280[1].

Screening investigations that should be performed in an acutely ill neonate suspected of having a genetic metabolic disorder

Urine

- Odour
- Dipstick tests for ketones, pH, sulphite (a)
- Reducing substances (testing for both glucose and non-glucose reducing substances)
- Amino, organic acid screens (including acylglycines)

Blood

- Full blood count/film
- Urea, electrolytes, anion gap, creatinine
- Glucose
- Calcium
- Blood gases
- Liver enzymes
- Uric acid
- Ammonium
- Lactate and pyruvate
- Amino acids (b)
- Carnitine and acylcarnitines (b)

Cerebrospinal Fluid

- Lactate and pyruvate
- Glucose
- Amino acids (b)

In the case of hypoglycaemia collect blood for the following when the child is hypoglycaemic

- Growth hormone
- Cortisol
- Insulin
- Free fatty acids
- ß – Hydroxybutyrate
- Acylcarnitine profile
- Urine should always be collected at the time of hypoglycaemia

(a) Sulphite is very labile. A negative test result does not exclude sulphite oxidase deficiency or the molybdenum cofactor defect.

(b) These tests should only be ordered after consultation with a biomedical geneticist or metabolic physician.

1. Christodoulou J, Wilcken B. Perimortem laboratory investigation of genetic metabolic disorders. *Semin Neonatol* 2004; **9**(4): 275-80.