YORK and SCARBOROUGH TEACHING HOSPITALS

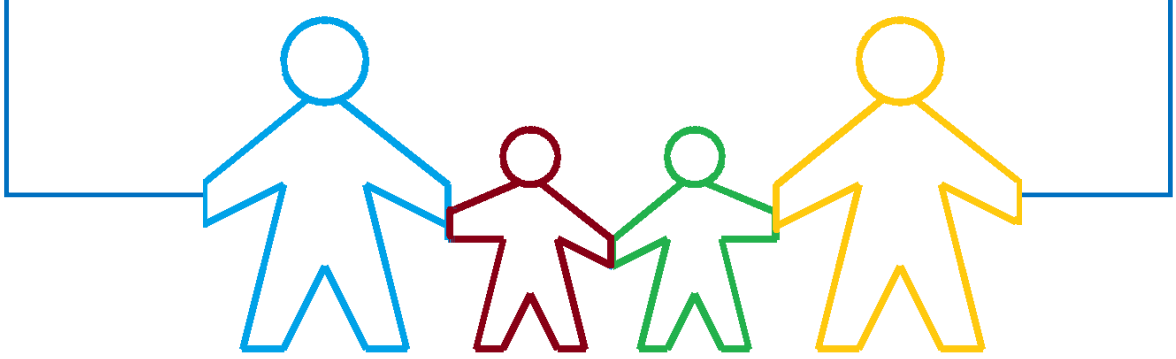
CLINICAL BIOCHEMISTRY

Part of Scarborough, Hull, York Pathology Services

S·H·Y·P·S

PAEDIATRIC HANDBOOK

DECEMBER 2024, VERSION 8.0



Contents

PAEDIATRIC CLINICAL BIOCHEMISTRY HANDBOOK

1. INTRODUCTION

This guide has been designed by the biochemistry department in consultation with the paediatric teams at York and Scarborough Hospitals. It is hoped that it will:

- Help you collect the correct specimen types for the tests you require
- Answer common questions about sample volumes and collection
- Prepare patients requiring specialist follow-up for referral to tertiary centres

If you have any remaining concerns or questions regarding sample volumes or types after reading this information, please contact the duty biochemist on extension 6366.

GENERAL INFORMATION

Biochemistry at York Hospital

The Laboratory Medicine Department reception is located on the ground floor at York Hospital with the departments on the first, second and third floors above. Our address is:

Laboratory Medicine Department,
York Hospital
Wigginton Road
YORK
YO31 8HE

The Laboratory Medicine Department is the block to the right of the main hospital entrance at Junction 2a. Biochemistry is situated on the second floor and is open from 08:30 to 17:30 Monday to Friday. Please see the 'contact details' section on page 4 for details regarding enquiries about results and specimens.

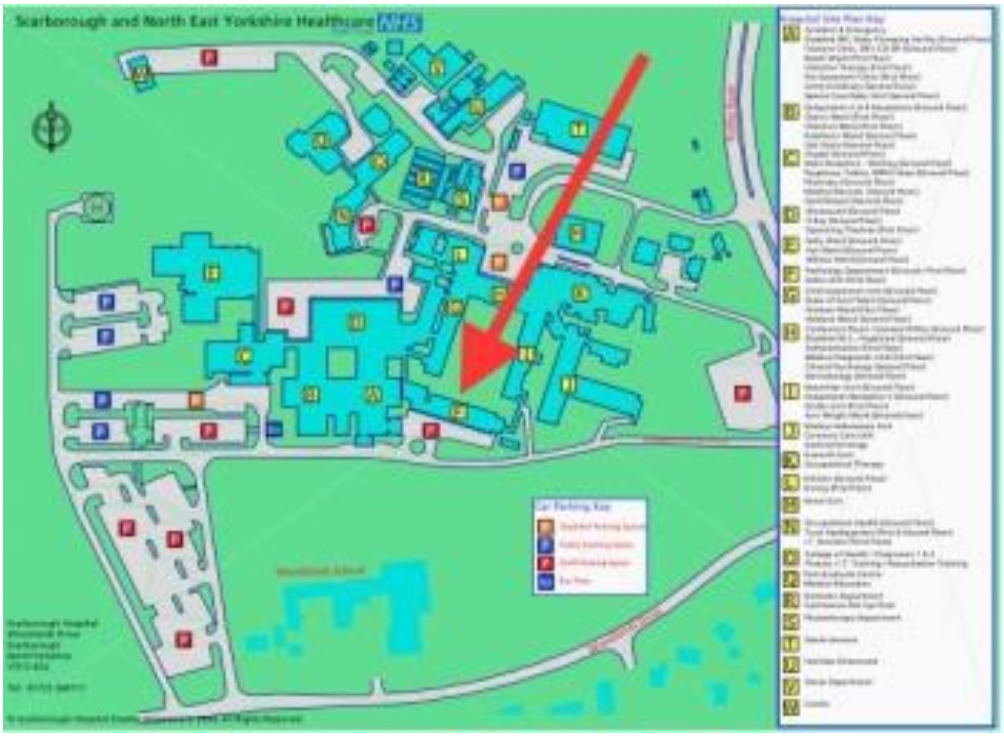
Biochemistry at Scarborough Hospital

The Laboratory Medicine Department is located in a purpose-built block at Scarborough Hospital. From inside the hospital and the South and North entrances, follow the signs to Pathology.

Our address is:

Laboratory Medicine Department,
Scarborough Hospital
Woodlands Drive
Scarborough
YO12 6QL

The department is labelled as point F on the hospital plan below:



The Pathology office, stores, phlebotomy, blood sciences and blood transfusion are on the ground floor. Access into the laboratory is restricted (electronically locked doors) and visitors are asked to report to the office reception. Please see the 'contact details' section below for details regarding enquiries about results and specimens.

CONTACT DETAILS

Please use the CPD reporting system whenever possible. This is available on the hospital network and is updated with completed results every 15 minutes.

For results & specimen enquiries:

YORK HOSPITAL
Blood Sciences Office, York (01904 72) 6802
Available 09:00 – 18:30 Monday to Friday

SCARBOROUGH HOSPITAL
Scarborough Reception (01723 34) 2351

Note that the York office cannot provide copy reports or updates on samples that are live within the laboratory at Scarborough. For copy reports or results on samples collected <24 hours prior, please use the details provided for Scarborough.

Out of hours enquiries:
On-call Biomedical Scientist Bleep via switchboard

The department operates a shift system and there is a biomedical scientist (BMS) in the hospital at all times. Outside routine hours only one BMS is on duty and they are often extremely busy. Please be patient and only bleep when absolutely necessary.

For clinical advice, specialist testing and interpretation of results:

Duty Biochemist (01904 72) 6366
Available between 09:00 and 17:00 Monday to Friday
Yhs-tr.biochemist@nhs.net

Out of hours clinical advice:

On-call Consultant Biochemist Contact via switchboard

Point of Care Testing:

Rachel Lampard (01904 72) 5890
Point of Care Co-ordinator

Mrs Maria de Ferrars (01904 72) 5599
Biochemistry lead for paediatrics
maria.deferrars@nhs.net

Dr. Deepak Chandrajay (01904 72) 5670
Consultant Chemical Pathologist and POCT lead
Deepak.chandrajay@nhs.net

Dr. Daniel Turnock (01904 72) 1847
Consultant Clinical Biochemist and Lead Clinician
daniel.turnock@nhs.net

For complaints and quality improvement:

Mr Carl Burkinshaw (01723 34) 2028
Network Biochemistry Manager
Carl.burkinshaw1@nhs.net

Mrs Emma Lovie
Chief Biomedical Scientist (01904 72) 4177
E.lovie@nhs.net

YORK HOSPITAL ONLY

For Phlebotomy and blood-taking enquiries:

Paediatric phlebotomy – Childrens' Assessment Unit (01904 72) 2018

SCARBOROUGH HOSPITAL ONLY

For Phlebotomy and blood-taking enquiries: (01723 34) 2652
Phlebotomy team
yhs-tr.PhlebotomyandClinicalSupportTeam@nhs.net

Deputy Phlebotomy manager - Scarborough (01723 34) 2048
Amy.provins@nhs.net

Scarborough Pathology Office: (01723 34) 2356

REQUESTS AND REPORTS

Any request form **must** display an **NHS number**, **date of birth, surname,** and **forename**, preferably printed in CAPITALS. Identifiers must be correctly spelt and complete, i.e. Initials are insufficient, as is an age of patient or just a year of birth.

Unlabelled samples are unfortunately not suitable for processing and will be discarded.

Where possible, please include all patient details including location, gender, and any relevant clinical information or drug history. This will ensure that results are delivered to the correct location with appropriate interpretive comments.

The table below shows the identifiers that may be included on the sample request form and sample tubes in order to process a request. The identifiers provided on the form and sample MUST MATCH e.g. samples should not be labeled 'Bob' if a form states the patient's name as 'Robert.'

	ESSENTIAL	Desirable	Optimal
Request Form	Tests required Surname Forename **AND** **At least one of the following**:- -NHS number - DOB -Hospital Number - A/E number	**Essential details** **AND** Requestor name Time and date collected Clinical Details Priority tests Urgency status* Risk status*	**Essential and desirable details** **AND** Patient's current location Patient's gender Patient's address Drug history Dose and Time for drug assays Consultant/GP contact details (essential for any immunology)
Sample Tube	Surname Forename **AND** **At least one of the following**:- -NHS number - DOB -Hospital Number - A/E number	**Essential details** **AND** Time and date collected Risk status*	**Essential and desirable details** **AND** Requestor name

*please see 'urgent or infectious samples' section overleaf for details.

URGENT OR INFECTIOUS SAMPLES

Urgent specimens should be taken by the doctor and hand-delivered to the laboratory immediately with a request form clearly marked URGENT.
The laboratory must be informed by telephone of the imminent arrival of an urgent sample **(For York: 6802, for Scarborough: (01723 34) 2351**, **York or Scarborough out of hours: bleep on call biomedical scientist).**

Samples marked "urgent" for which there has been no telephone call will be treated as routine.

High Risk Samples are those collected from a patient that has (or is suspected of having) any disease classed as category 3 or above, and any of the following:

HIV TB CJDv HepB HepC

All samples high risk samples should be taken by medical staff, **clearly labelled as high risk of infection with a yellow sticker, and double bagged.**
These samples CAN NOT be transported using the vacuum tube system.

1) *Requests made by Ordercomms electronic requesting*.
The High Risk box must be ticked. This ensures a subtle format change to the request form which, along with the use of double bagging, provides all the labelling required. Samples must also be labelled with a high risk sticker.

2) *Other requests*.
Where no Ordercomms requesting is available the infection risk of the patient must be indicated on the request card, in addition to a sticker on the sample. Samples should be double bagged (placed inside a second request form bag).

Any queries regarding high risk samples should be directed to the on-call microbiologist (via switchboard). If Phlebotomists are asked to take blood from patients at high risk of infection or being barrier nursed, they must be informed of the situation.

Requesting further tests on samples already received in the laboratory

We keep most of the samples we receive for approximately 3 days before they are discarded. Some samples are kept longer depending on the tests requested. If you want to add further tests to a sample please use the add-on functionality in CPD. If the desired test is unavailable, please ring (01904 72) 6802 OPTION 1 in York or the laboratory on (10723 34) 2351 in Scarborough.

We will check if we still have the sample and then add the tests. Please note that some analytes are labile and deteriorate rapidly. In these cases we will tell you that the sample we have left is unsuitable and fresh sample will have to be taken.

Reports

Most reports are available to view on CPD and completed reports are uploaded electronically every 15 minutes for the hospital.

Reference Ranges

Age- and sex-related reference ranges are printed on the report forms alongside the result, wherever these are in use. Please contact the duty biochemist if you have any queries regarding paediatric reference ranges (772 6366).

PHLEBOTOMY (BLOOD TAKING)

YORK HOSPITAL ONLY

Paediatric blood samples are collected on the Childrens' Assessment Unit (CAU) on Ward 18, or by a qualified healthcare professional on the paediatric ward. To arrange an appointment for sample collection, please contact CAU (01904 72 2108). Note that phlebotomy do not routinely bleed paediatric patients at York.

SCARBOROUGH HOSPITAL ONLY

The Scarborough phlebotomy team offer blood taking services to children aged 4 and above in the main outpatient area (reception B). Please contact Amy Provins (Deputy Phlebotomy Manager) to arrange an appointment for sample collection on 01723 342048 or via email (Amy.provins@nhs.net). Children under 4 years of age will need to contact Rainbow Ward to arrange sample collection within the hospital (Tel. 01723 362336).

PREPARATION

To avoid errors in sample collection, when bleeding paediatric patients, we ask that:

(1) You familiarise yourself with the requirements for your samples BEFORE bleeding a patient, using the information in section 3.
Please highlight any special requirements to the phlebotomist. If you remain unsure about requirements for your specific investigations, phone the duty biochemist (01904 72 6366). You may be directed to other departments if your query relates to haematology or immunology tests.

Lists of sample requirements provided by other hospitals may not necessarily be valid at your local lab. Additionally, some samples need to be delivered to the lab within a specific timeframe or under special conditions (e.g. on ice, or with specialist paperwork)to ensure accurate results. These are listed in the tube type tables in section 3.
If these requirements are not met, the sample may not be processed.

(2) Any samples requiring ice or immediate delivery to the lab are taken to the specimen reception hatch by hand.
This is the only way to guarantee safe delivery – air tubes may be unsuitable for some sample types and may fail, causing samples to be lost or delayed.

(3) You consider test priorities carefully before proceeding with phlebotomy and indicate any tests that are a priority on the sample request form.
This is particularly crucial if there is a limited sample volume or specialist test.

Specialist tests may need to be transported to referral laboratories in other parts of the UK. Where large numbers of specialist investigations are being carried out, each needing to be sent to a different centre, the number of tubes required may add up very quickly!

(4) No more than 4 tests are ordered to a single paediatric tube - please provide an additional sample when ordering 4 or more routine tests on the same sample type.
If this is not possible, consider splitting the number of requests across separate phlebotomy episodes. Please also note that certain tests may require a separate sample, as indicated in section 3.

(5) Adult sized tubes are used wherever possible – this avoids difficulties with sample volume and quality associated with using numerous small tubes.

2. TESTS AND TUBES

TURNAROUND TIMES AND TEST REPERTOIRE

We are able to measure over a hundred analytes in the laboratory. Most assays are performed daily but some specialist tests are performed less frequently or sent away to other laboratories. Sending samples to other laboratories may prolong the time taken to obtain results and require collection of extra samples (see section 3).

Turnaround times are calculated from the time that the sample is received in the laboratory to the result becoming available to the user in electronic format. Exact turnaround times for any test can be supplied on request, however, as a guide:

Test Type	Examples	50% reported within	75% reported within	95% reported within
Routine, non-urgent chemistry	CRP Glucose	45 minutes	1 hour	2.5 - 3 hours
Urgent Samples	Glucose marked 'urgent' & reception informed	1 hour		
Endocrine Tests	TFT, FT3, B12, ferritin			24 hours
	LH, FSH, Oestradiol, Testosterone			3 days

Test Repertoire

For simplicity the following tests are grouped with other, related tests:

Test Group	Analytes Included as Standard
Bone Profile	Calcium, Phosphate, Albumin, ALP (Alkaline phosphatase)
Liver Function Tests (LFT)	Bilirubin, Total Protein, Albumin, ALP, ALT (ALT = Alanine amino transferase)
Urea and Electrolytes (U&E)	Sodium, Potassium, Urea, Creatinine (eGFR is not valid in patients <18 years of age)

If you only require a single test or selected tests from a group please indicate which tests are required by writing their names in the space below the request boxes.

THIS IS ESPECIALLY IMPORTANT WITH LOW-VOLUME PAEDIATRIC SAMPLES when there may be insufficient sample to perform all the requested tests. Please highlight any priorities during sample analysis to ensure that any crucial tests are performed first.

Blood gases and ionised calcium

These are measured on the machines in SCBU and Delivery ward on both sites, with meters available in numerous other hospital locations if required. For a full list of meters available at each site, please see:

York and Scarborough Teaching Hospitals NHS Foundation Trust - Blood Gas Machine and Ketone Meter Locations (yorkhospitals.nhs.uk)

TUBE TYPES AND ORDERING

Sample Preservatives

All samples deteriorate from the time they are collected but this can be minimised by transporting samples to the laboratory as soon as possible and following any special requirements listed in section 3. The following samples are available for paediatrics:

Plain gel (serum): BROWN TOP →
Suitable for most routine biochemistry tests

Lithium Heparin (plasma): GREEN TOP →
Suitable for most routine biochemistry tests
Mandatory for some metabolic tests
e.g. very long chain fatty acids

EDTA (plasma): PURPLE TOP →
Required for routine haematology tests (e.g. FBC), as well as PTH and some specialist biochemistry.
NOT SUITABLE for trace metal analysis.
See tubes table for further details.

Fluoride Heparin (plasma): GREY TOP →
Mandatory for measurement of glucose and lactate

Sodium Citrate (plasma): BLUE TOP →
Used within haematology for clotting studies

Ordering Consumables

YORK HOSPITAL

There is a form for ordering consumables from Pathology Reception: This form can be sent into the laboratory or faxed to 01904 726358. Please order weekly and try not to keep large stocks of consumables as some items deteriorate with time.

Consumables Order Form.doc

SCARBOROUGH HOSPITAL

Consumables are ordered by filling in an order form, which can be obtained from Pathology stores (email below). The completed form should be scanned and emailed to pathologystores@york.nhs.uk or delivered by hand. Please allow 3-5 days for order processing. As some items deteriorate with time, we advise placing regular orders rather than keeping large stocks of consumables.

DYNAMIC FUNCTION TESTS (DFTs)

The most commonly performed dynamic function test in paediatric patients is the combined pituitary function test. Like all DFTs, this is complex in terms of patient preparation, sample collection and generation of reports in the laboratory.

As the samples collected are extremely precious, we recommend that you:

(1) Perform the test in the morning. As the protocol takes 3 full hours to complete, afternoon testing results in samples reaching the laboratory at the busiest time of day (late afternoon/evening) and they may not be processed efficiently.

(2) Familiarise yourself with the protocol and sample requirements before beginning the test. Collect and label all tubes required in advance.

(3) Hand deliver samples to the lab – **do NOT use the air tube**

(4) **Deliver all samples to the laboratory promptly once the final set has been collected from the patient (at 180 min).** Handling DFT samples is typically complex and delivery of samples for more than one patient at the same time greatly increases the chance of human error in the laboratory.

(5) Use the step-by step guide below when placing on order in CPD.

Ordering a Combined Pituitary Function Test in CPD

The combined pituitary function test MUST be carried out as specified in the protocols supplied by specialist healthcare professionals. This table provides an overview of the test and samples required:

Time Point	Test for	Tubes required
Preparation	Follow patient preparation instructions detailed in the clinical protocol. Collect and label any sample tubes required during the test.	
Baseline (0 mins)	**ACTH**	**1 x EDTA** (purple top) Deliver to lab immediately (within 30 min)
	EITHER testosterone OR oestrogen	1 x brown top **serum** Specify whether oestrogen or testosterone is required
	TSH, FT4, LH, FSH, prolactin, GH, IGF-1, cortisol	Minimum of 2 x **full tubes** (brown top **serum** or green **lithium heparin**) 1 x brown top **serum** for IGF-1
Give Glucagon as specified by clinical protocol		
30 mins	**Cortisol, Growth Hormone**	2 x full tubes (**serum** or **lithium heparin**)
60 mins	**Cortisol, Growth Hormone**	2 x full tubes (**serum** or **lithium heparin**)
90 mins	**Cortisol, Growth Hormone**	2 x full tubes (**serum** or **lithium heparin**)
120 mins	**Cortisol, Growth Hormone**	2 x full tubes (**serum** or **lithium heparin**)
150 mins	**Cortisol, Growth Hormone**	2 x full tubes (**serum** or **lithium heparin**)
180 mins	**Cortisol, Growth Hormone**	2 x full tubes (**serum** or **lithium heparin**)

To order in CPD:

- Go to the CPD "Place order" screen, select patient, consultant and enter clinical details as normal.
- Select BIOACTH from a list of pathology order items as you would for a normal blood test order.

- In the next field, select either BIOTES (testosterone) or BIOOES (oestradiol) depending on whether the patient is male or female.
- Click "Order sets" button and search "pituitary" to find the order set called "**PITUITARY FUNCTION TEST – PA**." This should add a list of items beginning with BIOPFT0 and ending with BIOPFT180. **If you can't find this order set, click the option for: "View sets I am not authorised to submit" and search again.**
- BEFORE clicking the "process order" button, the "Sample Sep" box on the right hand side should be ticked for each item on the list. This will ensure that 9 separate request forms are printed.
- Clicking 'process order' will bring up a prompt to send the ACTH sample to the laboratory immediately (all other samples can be stored on the ward).
- Once all forms are printed, collect the ACTH sample and handwrite the time of collection on the request form. Deliver this to the laboratory immediately by hand (must arrive within 30 minutes of sample collection).
- Follow the clinical protocol for collecting the rest of the samples. Label each sample taken during the test with the actual time of collection (e.g. 10:27 if sample collected at this time) AND the time in relation to the DFT (e.g. t = 0, t = 30 min etc). Store samples and forms on the ward until the end of the test.
- Once test is complete and all blood samples have been collected, CHECK ALL TIMES AND PATIENT INFORMATION on the samples and request forms to ensure that these match up. Send all seven samples down to the laboratory.

Please refer to the Endocrine Protocols or contact the duty biochemist (01904 726366) for further information or assistance.

SWEAT TESTING FOR CYSTIC FIBROSIS

This involves electrical stimulation of sweat glands near the skin surface and collection of a sweat sample by a trained member of laboratory staff. Patients should be >2 weeks of age (preferably >3 months), well hydrated and without systemic illness.

Sweat testing may need to be rescheduled:

- if an insufficient volume is collected (e.g. patient very young or dehydrated)

- if the collection site is eczematous

- if there is evidence of systemic illness or oedema

- in patients on topimarate, 9-alpha fludrocortisone, or open delivery oxygen systems

(Guidelines for the Performance of the Sweat Test for the Investigation of Cystic Fibrosis in the UK v.2)

To arrange an appointment for sweat testing:

YORK HOSPITAL

Please phone 5802 to arrange an appointment (WEEKDAYS and typically >7 days in advance).

SCARBOROUGH HOSPITAL

Contact Alan Shepherd, senior biomedical scientist (01723 345372 / alan.shepherd3@nhs.net) or the paediatric admin team (childhealthadmin@nhs.net) to arrange. As the laboratory needs to ensure that a trained tester is available, we advise booking at the earliest possible opportunity.

CSF NEUROTRANSMITTERS
Monoamine metabolites, Pterins, Methyl Tetrahydrofolate and Pyridoxal-5-phosphate (Vitamin B6)

These are highly unstable analytes, which are collected by lumbar puncture into a special preservative, and must be collected onto ice by a member of laboratory staff at the patient bedside. If this investigation is planned, the laboratory should be contacted at the earliest possible opportunity so that specialist collection tubes can be ordered and arrangements made for a trained member of staff to be available.

Failure to notify the laboratory promptly may result in appointment cancellation.

Ideally, one of the consultant biochemists should be copied into the letter stating the date and time of the lumbar puncture appointment. This should be confirmed by contacting the duty biochemist on 6366 a week before the appointment takes place.

A specialist request form should be completed prior to lumbar puncture – this can be obtained by contacting the duty biochemist or accessing:

csf-neurotransmitter-request-form.pdf (uhnmpathology.com)

GENETIC TESTING
(Microarray, Karyotyping and Molecular Genetics)

Genetic test requests should generally be made through the Yorkshire Regional Genetics service. Referral to a clinical geneticist ensures that patients are appropriately counselled prior to testing, that a targeted test or panel of gene tests are carried out, and that relevant ethical guidance is followed.

Where necessary, samples may be referred to Leeds on behalf of the genetics team or an experienced specialist (usually a consultant). These requests MUST be accompanied by a fully completed genetics request card, which can be accessed using the links below. Please also note that DNA testing requires a separate sample to any other tests to avoid any possibility of contamination during routine analysis.

- For **most molecular genetic or cytogenetic tests** (microarray, karyotyping, genes or gene panels):

  411.027-Rare-Disease-Referral-Form-v3.0web.pdf (ney-genomics.org.uk)

- For DNA testing in suspected **mitochondrial disorders**:

  Newcastle-Referral-form.pdf (newcastle-mitochondria.com)

DRIED BLOOD SPOT SCREENING FOR FABRY DISEASE IN FEMALE PATIENTS

Fabry disease is a lysosomal storage disorder, and can be detected on a white cell enzyme panel or using a dried bloodspot (a more targeted method if there is strong clinical suspiscion of Fabry's).

If the dried bloodspot method is used, abnormal results in female patients are followed up using genetic testing.

For this reason, **ANY female patient undergoing Fabry's screening using the dried blood spot method should be made aware that genetic testing is a possibility and asked to provide consent at the outset.**

When consent has been obtained, this should be written onto the request form that comes with the sample i.e. "Consent for genetic testing obtained."

Failure to document that consent has been obtained may result in a delayed diagnosis as the referral laboratory that carries out testing for Fabry's disease will not proceed with genetic follow-up unless they have a clear, written statement of consent.

Consent is not necessary if a full white cell enzyme screen is being performed, as genetic testing for Fabry's does not form part of this screening procedure (any abnormal findings on the white cell enzyme screen can be followed up using more specific tests, such as the dried blood spot method. It is only at this point that the possibility of genetic testing will need to be considered).

GENERAL GUIDANCE FOR INTERPRETING BIOCHEMISTRY RESULTS IN PAEDIATRICS

This topic is the subject of an excellent review in the British Medical Journal (see https://www.bmj.com/content/bmj/361/bmj.k1950.full.pdf)

Although the laboratory supplies paediatric reference ranges wherever possible, these are not available for every test offered by the laboratory. This may be because:

- Concentrations of an analyte in a particular age group have not been studied due to challenges in sampling and analysis (e.g. premature neonates)
- The evidence base for paediatric reference ranges in tests which have been recently been developed is poorly established (e.g. Faecal Calprotectin)
- Concentrations of an analyte vary over a huge range during early life and it is not practical to include a full list of paediatric ranges in tests which are more commonly requested in adults (e.g. sex hormones, gonadotrophins).
- Published ranges cannot always be used interchangeably between different laboratory methodologies or analytical platforms

As such, you should be aware that tests results may sometimes be flagged as 'abnormal' in a child due to an adult range having been applied and interpret all values in the context of a patient's clinical presentation and developmental stage.

If you require assistance with result interpretation, please contact the Duty Biochemist on 01904 726366.

We will do our best to help you source an appropriate reference range, but please be aware that we are limited by what has been published in the scientific literature.

When ordering a specific test, it is essential to remember that you are committing to the responsibility of acting on the end result and should have a reasonable idea of how the result that is returned to you will influence patient management.

3. TABLES OF SAMPLE REQUIREMENTS

ROUTINE BIOCHEMICAL TESTS

The laboratory will make every attempt to ensure that the tests required are carried out on the sample volume provided, but **please be aware that multiple tests on the same sample type may require multiple tubes. If possible, provide a second sample where there are 4 or more routine tests to perform. A new tube is also required for any tests requiring a 'separate sample' where indicated below.**

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Alanine aminotransferase (ALT) ***Part of LFT profile*** Increases indicate liver cell damage.	**Serum** or **Lithium Heparin**	Daily	F = 0-34 IU/L M = 0-45 IU/L
Albumin - *Part of LFT and Bone Profiles* Low levels reflect kidney/GI loss, infection, malnutrition, hemodilution or redistribution	**Serum** or **Lithium Heparin**	Daily	<1 year: 30-45 g/L 1-16 years: 30-50 g/L
Alkaline Phosphatase (ALP) ***Part of LFT and Bone Profiles*** Elevated in bone and liver disease or benign transient hyperphosphatasia.	**Serum** or **Lithium Heparin**	Daily	Neonate: 70-380 U/L Child <16: 60-425 U/L >16 years: 30-130 U/L
Alpha Fetoprotein (AFP) Produced by the foetal liver and yolk sac during gestation and up to 1 year of age. Used alongside other tests to diagnose and monitor hepatoblastoma, and in detection of interuterine disease	1mL blood in **Serum** or **Lithium Heparin**	Daily	>1 year-old: <7 kU/L Please contact the duty biochemist to discuss age-specific ranges in children <1 year old.
Ammonia Investigation of suspected hyperammonaemia e.g. in encephalopathy, hyperventilation and acid base disturbances.	1mL in **EDTA** **Samples must arrive within 30 minutes of collection (or 60 minutes on ice).**	Daily	**Group** **umol/L** Sick/premature: <150 Neonate: <100 Child <16 years: <50
Aspartate aminotransferase (AST) Raised in liver and muscle damage. N.B. this is NOT included in the LFT profile.	**Serum** or **Lithium Heparin**	Daily	F = 0-31 IU/L M = 0-35 IU/L
Bicarbonate (CPD code = BIOSBIC) Informative about acid/base status.	**Serum** 1.3mL	Daily	22-29 mmol/L
Bilirubin - *Part of LFT profile* Total bilirubin may be raised for numerous reasons, including 'breast milk jaundice,' haemolysis, or hepatocellular dysfunction	**Serum** or **Lithium Heparin** **Samples should be protected from light if possible.**	Daily	14 days-16 years: <21 umol/L
Bilirubin Split (Conjugated and Unconjugated). CPD code = BIODBIL Differential diagnosis of jaundice. Unconjugated bilirubin predominates in haemolytic or 'breast milk' jaundice; conjugated bilirubinaemia indicates other causes (e.g. infection, biliary atresia, IMD).		Daily	Direct bilirubin >33% of total is generally considered conjugated hyperbilirubinaemia.
Bone Profile ***Includes total and adjusted calcium, phosphate, albumin, total protein, ALP***	**Serum** or **Lithium Heparin**	Daily	See individual analytes

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Calcium (Total) - *Part of Bone Profile* Assessment of calcium homeostasis. Adjustment provides an estimate of free calcium, based on total calcium and albumin concentrations.	**Serum** or **Lithium Heparin**	Daily	Neonate: 2.0-2.7 mmol/L Child <16: 2.2-2.70 mmol/L
Ceruloplasmin Copper binding serum protein used to screen for Wilson's disease	**Serum** or **Lithium Heparin** Separate sample required.	Weekly	0.20-0.60 g/L
Chloride Note routinely reported, but may be useful in investigation of electrolyte imbalance and acid-base disorders.	**Serum** or **Lithium Heparin**	Daily	95-108 mmol/L
Cholesterol Diagnosis and monitoring of disorders of lipid metabolism, such as familial hypercholesterolaemia or diabetes.	**Serum** or **Lithium Heparin**	Daily	Levels >6.5 mmol/L in a child <16 years of age may be suggestive of FH.
LDL Cholesterol Calculated from total cholesterol and triglyceride in fasting sample.	**Serum** or **Lithium Heparin**	Daily	Levels >4.0 mmol/L in a child <16 years of age may be suggestive of FH.
C-reactive protein (CRP) Acute phase reactant, particularly increased in bacterial infection	**Serum** or **Lithium Heparin**	Daily	<5 mg/L
Creatinine ***Part of U&E profile*** Measurement of renal function. Affected by muscle mass, muscle breakdown and protein intake as well as glomerular function. For this reason, age-related ranges are a guide only.	**Serum** or **Lithium Heparin** **Please specify if dopaminergic drugs or vitamin supplements are used.**	Daily	Dependent on muscle mass, roughly (umol/L): Neonate: 22-90 Infant <1 yr: 11-34 Child <14 years: 21-65 Age>14 years: 49-104
Creatine kinase (CK) Non-specific indicator of muscle damage. Raised in inherited myopathies and neuromuscular disorders, such as Duchenne Muscular Dystrophy.	**Serum** or **Lithium Heparin**	Daily	**No-specific paediatric range – as a guide, adult ranges are:** F = 25-200 IU/L M = 40-320 IU/L
Gamma glutamyl transpeptidase (GGT) Sensitive indicator of liver disease. Increased after exposure to enzyme inducing drugs (e.g. ethanol) or hepatobiliary damage. N.B. this is NOT included in the LFT profile.	**Serum** or **Lithium Heparin**	Daily	F = <38 U/L M = <55 U/L
Glucose Diagnosis and monitoring of diabetes mellitus and hypoglycaemia.	**Fluoride Heparin**	Daily	Fasting: 2.5-6.0 mmol/L
Glucose (CSF) Measured in bacterial meningitis	1mL CSF in **Fluoride Heparin**	Daily	0-2 years: 2.2-3.9 mmol/L >2 years: 3.3-4.4 mmol/L
Haemoglobin A1c Monitoring of glycaemic control in diabetes mellitus – **NOT suitable for diagnosis** of diabetes in children.	**EDTA** 1.3mL	Daily	20-42 mmol/mol

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Haptoglobins Decreased with *in vivo* haemolysis or ineffective erythropoiesis. Increased after corticosteroids and acute phase response.	**Serum** or **Lithium Heparin** Separate sample required.	Weekly	0.30-2.00 g/L
Iron Used in assessing iron toxicity. Ferritin is a better indicator of iron storage.	**Serum** or **Lithium Heparin** Please include details of any iron therapy.	Daily	0-2 years: 9-21 umol/L M >2 y: 12-30 umol/L F >2y: 9-27 umol/L
% Iron saturation Calculates % of transferrin bound to iron.		Daily	15-50%
Lactate (CSF). CPD code = BIOCLAC Increased in some inherited metabolic disorders (e.g. mitochondrial cytopathy) also bacterial and fungal meningitis	1 mL CSF in **Fluoride Heparin**	Daily	1.1-2.4 mmol/L
Lactate (whole blood) Investigation of hypoglycaemia, inborn errors of metabolism or unexplained acidosis.	1mL in **Fluoride Heparin** Patient should be resting.	Daily	0.6-2.5 mmol/L
Lactate Dehydrogenase (LDH, CPD code = BIOLDH1) Measured in jaundice and suspected haemolysis. Also suitable for use in megaloblastic and pernicious anaemias, leukaemia, lymphomas, and liver disease	**Serum** or **Lithium Heparin**	Daily	<248 IU/L
Liver Function Tests (LFT) ***Includes ALT, ALP, Total Protein, Albumin, total bilirubin.***	**Serum** or **Lithium Heparin**	Daily	See individual analytes
Magnesium Measured in hypocalcaemia (low Mg impairs PTH release), nutritional deficiency, GI losses (e.g. severe diarrhoea) and renal tubulopathy. Serum Mg levels are not a good indicator of body stores.	**Serum** or **Lithium Heparin**	Daily	Neonate: 0.6-1.0 mmo/L Child: 0.7-1.0 mmol/L
Osmolality (serum) Measurement of the total osmotically active particles in serum. Comparison with the calculated osmolarity can show if there is an "osmolar gap" due to unidentified substances in the serum (e.g. ketones).	**Serum**	Daily	275-295 mmol/kg (see specialist protocol for interpretation in water deprivation tests)
Phosphate ***Part of bone Profile*** Investigation of hyper- or hypo-phosphataemia, rickets, bone and renal disorders. Increases with standing or delayed separation from red cells	**Serum** or **Lithium Heparin**	Daily	**Age** **mmol/L** Neonate: 1.3-2.6 Infant <1 yr: 1.3-2.4 1-16 years: 0.9-1.8
Potassium ***Part of U&E Profile*** Monitored in renal disorders and infants requiring nutritional support. Old or haemolysed samples are not measured due to the spurious rise in potassium from the lysed red cells.	**Serum** or **Lithium Heparin** Samples **MUST** show a time of collection	Daily	**Age** **mmol/L** Neonate: 3.4 - 6.0 Infant <1 yr: 3.5 - 5.7 1-16 years: 3.5 - 5.0

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Protein (CSF) CPD code = BIOCPRO. May be increased in meningitis and CNS tumours	Plain CSF	Daily	0-1 week: 0.45-1.09 g/L 1-4 weeks: 0.34-0.98 g/L 1-4 m: 0.19-0.71 g/L 4-6 m: 0.21-0.37 g/L 6m–1y: 0.13-0.41 g/L 1-16y: 0.12-0.32 g/L
Sodium (serum) - *Part of U&E profile* Main use is as a measure of the state of hydration of a patient.	**Serum** or **Lithium Heparin**	Daily	133-146 mmol/L
Transferrin Measured to estimate iron saturation in the investigation of iron overload and also as a marker of nutritional status	**Serum** or **Lithium Heparin** Please include details of any iron therapy.	Daily	2.0-3.6 g/L
Triglycerides Diagnosis and monitoring of disorders of lipid metabolism, such as obesity and diabetes. Levels may be falsely raised due to contamination with glycerol in nappy creams or glycerol kinase deficiency.	**Serum** or **Lithium Heparin** Fasting sample preferred – please specify	Daily	<1.7 mmol/L
Urea and Electrolytes (U&E) ***Includes sodium, potassium, urea, creatinine.*** eGFR is not valid in patients under the age of 18 years old.	**Serum** or **Lithium Heparin** An accurate collection time must be provded.	Daily	See individual analytes
Urea - *Part of U&E profile* Measure of renal function (with creatinine) and degree of hydration (with sodium). Also affected by protein intake, GI bleeding and liver function.	**Serum** or **Lithium Heparin**	Daily	Neonate: 0.8-5.5 mmol/L Infant: 1.0-5.5 mmol/L 1-16 y: 2.5-6.5 mmol/L
Valproate Only useful to check compliance or possible overdose, little use for therapeutic monitoring (clinical effects more reliable).	**Serum** or **Lithium Heparin** collected **before** dose taken	Daily	No well-established range. As a rough guide: 50-100 mg/L
Zinc – *Part of Trace Elements Profile* Estimation of the zinc nutritional state of a patient. Must be interpreted with the serum albumin level (as some zinc is bound to albumin) and CRP (infection and inflammation decrease serum zinc).	**Serum** or **Lithium Heparin** Separate sample required.	Weekly	9.8 - 17.9 umol/L

IMMUNOASSAY AND ENDOCRINE TESTS

This includes most endocrine tests and haematinics. Please note that although plasma samples are now acceptable for the majority of these tests, testosterone remains an exception as different tube types can give significantly different results.

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Adrenocorticotrophin (ACTH) Second line test for adrenal & pituitary disorders.	1mL blood in **EDTA** **MUST be put on ice and transported to the lab within 30 minutes of collection.** Requires a separate sample to any other EDTA tests.	Sent away (Hull)	<47 ng/L **Requires a paired cortisol for interpretation.**
Aldosterone Investigation of hypokalaemia / hypertension	0.3mL **Serum** or **Lithium Heparin** Please include medication details. Requires a separate sample to any other tests.	Sent away (Leeds)	See report; interpreted in light of renin result
Anti-Mullerian Hormone (AMH) Investigation of ambiguous genitalia, assessment of ovarian reserve and testicular function in patients at risk of infertility (e.g. Turners, Klinefelters)	1mL blood in **EDTA** or **Lithium Heparin** to arrive at the lab a.s.a.p (sample must be processed on the day of collection)	Sent away (London)	See report for age- and gender related reference ranges
Androstenedione Measured with DHAS in adrenal androgen disorders.	**Serum only** Send separate sample to any other serum tests.	Sent away (Hull)	0.8 - 4.7 nmol/L Please contact the duty biochemist to discuss ranges for a particular age or pubertal stage
Cortisol (blood 9 a.m) Investigation of adrenal cortical function. Hydrocortisone and prednisolone interfere.	**Serum** or **Lithium Heparin** **Please include details of any steroid medications or dynamic function tests and record TIME of collection.**	Daily	150-650 nmol/L (a.m)
Dehydroepiandrosterone (DHAS) Measured with androstenedione in adrenal androgen disorders.	1.3mL **Serum** Send separate sample to any other serum tests.	Sent away (Hull)	0.5 – 10.6 umol/L Please contact the duty biochemist to discuss ranges for a particular age or pubertal stage
Ferritin To detect iron deficiency and iron overload (in rare cases). Increases in the acute phase, so CRP also measured.	**Serum** or **Lithium Heparin** **Please specify if recently transfused or taking iron supplements.**	Daily	M = 22-322 ng/mL F = 20-291 ng/mL
Folate (serum) Investigation of folate status.	**Serum** or **Lithium Heparin**	Daily	>3.4 ug/L
FSH Assessment of precocious or delayed puberty, often as part of stimulation or suppression tests.	**Serum** or **Lithium Heparin**	Daily	Pre-pubertal: <1 IU/L

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Growth Hormone Investigation of hypoglycaemia, short stature and gigantism. Secretion is best assessed by stimulatory or suppression tests.	**Serum** or **Lithium Heparin** **N.B. random levels provide little information, as secretion is pulsatile (low levels may reflect a missed pulse).** Separate sample recommended.	Weekly	N/A – levels are best interpreted as part of stimulation or suppression tests (see individual protocols).
17 Hydroxyprogesterone Diagnosis of CAH due to 21 hydroxylase deficiency in precocious puberty/hirsuitism.	0.5 mL **Serum** Collect samples prior to dose if on treatment for CAH. Requires a separate sample	Sent away (Leeds)	0-5 days: 0-3 nmol/L 5 days -16y: 0-4 nmol/L
Insulin (and C-peptide) Investigation of hypoglycaemia and newly-diagnosed diabetes. For hypoglycaemia, glucose must be measured simultaneously using the laboratory assay.	**Serum only** **Samples must be transported ON ICE and arrive at the lab within half an hour of collection.** Send separate sample to any other serum tests. **Also send sample for glucose if screening for hypoglycaemia.**	Sent away (Guildford)	See report – results are interpreted in light of blood glucose level at the time of sampling. **Lab glucose must be <2.2 mmol/L for hypoglycaemia screening**
Insulin-Like Growth Factor I (IGF-I) Useful in the investigation of growth disorders.	**Serum only.** Separate sample required to any other serum tests.	Weekly	See report for exact age-related range.
LH Assessment of precocious or delayed puberty, often as part of stimulation or suppression tests.	**Serum** or **Lithium Heparin**	Daily	Pre-pubertal: <2 IU/L
Oestradiol Assessment of delayed or precocious puberty in females and disorders of sexual development.	**Serum** or **Lithium Heparin**	Daily	Male: 0 – 206 pmol/L Female Pre-menstrual: 50-150 pmol/L Female Post-menarche: 150 -1500 pmol/L
Parathyroid Hormone (PTH, CPD code = BIOPTH1) Useful in clarifying the cause of hyper- and hypo-calcaemia. Interpretation should be made in light of the serum calcium level.	**EDTA** **Please send to the laboratory within 24 hours of calcium measurement.** Separate sample preferred i.e. 2 EDTA samples for PTH and FBC.	Daily	Levels must be interpreted in context of serum calcium. For normal calcium: 1.2-8.5 pmol/L
Renin Measured in the investigation of causes of juvenile hypertension (suspected primary hyperaldosteronism)	**Lithium Heparin** **Must be transported at room temperature and arrive at the lab within 30min**. Please include medication details. Separate sample required.	Sent away (Leeds)	0.5-3.5 nmol/L/h random sample – interpret in the context of UE and aldosterone results.
Sex hormone binding globulin (SHBG) Assessment of free androgen index. Reduced in obesity and thyroid dysfunction.	**Serum** or **Lithium Heparin**	Daily	F = 18-114 nmol/L M = 10-57 nmol/L

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
T3 (free) Investigation of hyperthyroidism.	**Serum** or **Lithium Heparin** Please include details of thyroxine therapy and any other medication.	Daily	3.5-6.5 pmol/L
T4 (free) Investigation of possible hyper- or hypothyroidism in conjunction with TSH		Daily	9-23 pmol/L
Testosterone Assessment of delayed or precocious puberty in males and disorders of sexual development. **N.B. all testosterone requests will be processed using a local immunoassay method in the first instance** – if specialist investigations are being undertaken, results are unusual or there is a discrepancy between clinical and laboratory findings, contact the duty biochemist to discuss **mass spectrometry.**	**Serum only - due to the prevalence of cross reacting steroids in infancy and childhood, we recommend contacting the duty biochemist (x6366) to arrange referral for a mass spectrometry assay where testosterone is a key diagnostic investigation.** Please ensure time of collection is included and aim to sample at 9am to avoid changes due to diurnal variation.	Daily (Mass spectrometry assay = weekly at Hull)	nmol/L F <12y: 0.07-0.69 M <1y: 0.42-0.72 M 1-6y: 0.1-1.12 M 7-12y: 0.1-2.37 M 13-17y: 0.98-38.5
TSH Raised in primary hypothyroidism	**Serum** or **Lithium Heparin** Please include details of thyroxine therapy.	Daily	0.55 - 4.78 mU/L
TSH (Down Syndrome ONLY, CPD code = BIODTSH.) Investigation of thyroid disorders in patients where venous sampling with a needle is not possible.	**Guthrie card** (with special label) Finger prick blood spots, leave to dry for 4h before packaging.	Sent away (Leeds)	<5 mU/L
Urine steroid profile	Please see 'Urine Tests' table on page 29		
Vitamin B12 Megaloblastic anaemia, dietary deficiency, malabsorption and some inherited metabolic disorders. Reference ranges are a guide, please interpret in the context of haematological / neurological findings	**Serum** or **Lithium Heparin** Please contact the duty biochemist to discuss testing if an inborn error of metabolism is suspected.	Daily	211-911 ng/L
Vitamin D Investigation of unexplained hypo- and hypercalcaemia, assessment of patients at risk of fat-soluble vitamin deficiencies.	**Serum** or **Lithium Heparin**	Daily	Deficiency: <30 nmol/L Insufficiency: 30-50 nmol/L

ALLERGY AND IMMUNOLOGY TESTS

This includes common requests in patients with symptoms of allergy, autoimmune disease or immunodeficiency. Further information on how to request individual allergy tests or test panels can be found in the text below this table.

ANALYTE (TEST SET)	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Alpha-1-antitrypsin Investigation of liver disease	2 mL **Serum.** Separate sample required to all other serum tests.	Weekly	See report for age related ranges.
Cryoglobulin Investigation of suspected Raynaud's disease, pain and numbness in fingers and toes. ESSENTIAL that samples are kept at 37°C.	2mL **Serum only.** **Contact the duty biochemist (6366) to arrange testing. Blood MUST be collected into a warmed flask supplied by the laboratory between 9am – 12 noon.**	Daily	Negative
C1 Esterase inhibitor Investigation of angioedema and low C4 complement.	**Serum only** Send separate sample	Sent away (Hull)	0.21 - 0.39 g/L See report for further details
C3 and C4 Complement Low in diseases such as SLE, nephritis, vasculitis, rheumatism and hereditary angioedema.	**Serum** or **Lithium Heparin** Separate sample required.	Weekly	C3: 0.90-1.8 g/L C4: 0.1-0.4 g/L
Electrophoresis (serum) Suspected immunodeficiency.	**Serum only** Please provide clinical details.	Daily	Normal pattern
Immunoglobulins ***Includes Ig A, Ig G, IgM*** Autoimmune disorders, chronic or recurrent infections	**Serum** or **Lithium Heparin**	Daily	Age related up to 9y for IgG and IgA and up to 6y for IgM. See report
Immunoglobulins IgG subclasses (CPD code = BIOGSUB) Persistent infection with apparently normal total immunoglobulin levels.	2mL **Serum** or **Lithium Heparin** A separate tube is required for this test.	Sent away (Sheffield)	Age related ranges. Please see report for range applicable to your age group.
Total IgE Allergic and atopic diseases; Please see text below for further information.	1 mL **Serum** – please note that requests for 'IgE' will give a total level ONLY. Requests for Ig E levels to specific allergens should be listed clearly and separately (see below)	Twice weekly	0-3 m: <5 KU/L 3-12m: <11 KU/L 1-5y :<29KU/L 5-10y: <52 KU/L 10-15y: <63 KU/L 15y – adult: <75 KU/L
Specific Ig E Allergic and atopic diseases; Please see text below for further information on ordering / link to specialist request form.	1 mL **Serum** - if >8 tests are selected or any tests are NOT offered at York (see next page) please collect an extra sample. **Please list specific IgE tests clearly and legibly on request form in BLOCK CAPITALS.**	Twice weekly (in house repertoire) or referred (Sheffield)	<0.35 kAU/L Please see individual reports for full interpretive guidance.
Tryptase Investigation of anaphylaxis (e.g. reactions to anaesthetic) **Take blood on presentation, then at 3 and 24hrs post-reaction.**	**Serum** samples taken on presentation and a 3 and 24-hours post-reaction. **Samples should arrive within 3 hours of collection.** Requires a separate sample.	Sent away (Leeds)	2-14 ng/mL

Allergy Testing and Specific IgE analysis

We test for most common allergens by measuring specific IgE to the suspected allergen. Panels are available in patients with typical allergy symptoms – these are:

Panel	Tests Included
Inhalent	Timothy grass, Alternaria alternata, Cladosporium herbarum, Birch and Mugwort.
Tree	Alder, Silver Birch, Hazel, Oak and Willow.
Mould	Penicillium chrysogenum, Cladosporium herbarum, Candida albicans, Aspergillus fumigatus, Alternaria alternata, Setomelanomma rostrata.
Weed	Ox-eye daisy, dandelion, plantain, golden-rod and Lamb's quarters.
Rodent	Guinea pig, Hamster, Rabbit, Rat and Mouse
Feather	Goose, Chicken, Duck and Turkey.
Food	Milk, egg, cod, wheat, peanut and soybean.
Fish	Cod, tuna, salmon, blue mussel and shrimp.
Mixed Nut	Hazel, Brazil, Almond, Peanut and Coconut.
Grain	Wheat, Rye, Barley, Rice
Caged bird feathers	Budgie, Canary, Parakeet, Parrot, Finch

A full lists of the tests offered at York and guide to interpretation can be found here: https://www.yorkhospitals.nhs.uk/our-services/a-z-of-services/lab-med/general-information/information-for-health-care-professionals1/advice-for-primary-or-secondary-care-clinical-biochemistry/

Please ensure that a detailed history is taken and that requests for testing to specific allergens are relevant and clearly legible on the request form (e.g. Ig E to WALNUT) as vague requests for 'IgE' will result in a total Ig E result without any further testing. You may also wish to use the laboratory's dedicated allergy request form (click below or contact the Duty Biochemist):

CB-TEM-ALLERGYRE Q v2.docx

Requests for less common allergens are sent to a reference laboratory, and a full range is available on request (contact duty biochemists on: 01904 72 6366). If extensive or unusual allergy testing is required (i.e. samples referred elsewhere for analysis), it is likely that more than one serum sample will be needed - **as a rough guide, please send (an) additional tube(s) where >8 specific Ig E tests are requested.**

In the event of any queries or requirement for clinical advice, please contact:

Hull Immunology Secretaries: 01482 461312.

Dr Anna McHugh
(Consultant Immunologist, Hull Royal Infirmary):
Tel: 01482 607710 / Email: anna.mchugh@nhs.net

SPECIALIST AND METABOLIC INVESTIGATIONS

The table below lists investigations for patients with symptoms of an inherited metabolic disorder (e.g. hypoglycaemia, seizures, cardiomyopathy or global developmental delay), genetic disorder, renal calculi, nutritional deficiency, or heavy metal poisoning. Please see 'urine tests' for any urine analyte (including metals, citrate, oxalate, urine organic acids and amino acids).

If the test you require is not listed below, contact the duty biochemist (01904 726366).

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Amino Acids Diagnosis of disorders of amino acid metabolism. **Please state whether a specific disorder is suspected.**	0.2 mL in **Serum** or **Lithium Heparin** Separate sample required.	Sent away (Leeds)	See report for specific pattern and interpretation.
Batten's Screen (palmitoyl protein thioesterase / PPT and tripeptidyl peptidase 1 / TPP) Investigation of progressive neurometabolic symptoms (seizures, vision loss, developmental regression)	5 mL whole blood in **EDTA** **Must be collected on Monday-Wednesday ONLY to avoid transport issues over the weekend.** Separate sample required.	Sent away (Manchest-er)	See individual report
Beta hydroxybutyrate (ketones) and Free fatty acids. Investigation of hypoglycaemia, diabetic ketoacidosis.	0.5 mL in **Fluoride Heparin**. Separate sample required. Measure alongside fatty acids and glucose.	Sent away (Sheffield)	Results are interpreted in light of glucose and free fatty acid concentrations.
Biotinidase (CPD code = BIOBIO) Enzyme involved in recycling biotin. Deficiency results in recurrent infection, developmental regression hair loss, skin rashes, visual and hearing defects.	0.2 mL in **Serum** **Samples must be delivered to laboratory immediately.** Separate sample required.	Sent away (Leeds)	4.4-12.0 nmol/min/mL
Calculus (Renal Stone analysis) Identification of stone components	Dry renal stone specimen	Sent away (London)	N/A
Carnitine and acylcarnitine Co-factors involved in free fatty acid and acyl CoA transport across membranes. Deficient in some errors of fatty acid oxidation with hypoglycaemia and cardiomyopathy (e.g. MCADD)	2 dried blood spots on a guthrie card(available from children's clinic - please leave to dry for 4h before packaging.)	Sent away (Leeds)	See report - specific pattern of metabolites may be either normal or suggestive of a particular disease.
CSF Neurotransmitters* (Monoamine metabolites, pterins, Vitamin B6). Investigation of cyclical seizures, and neurological disorders.	**Testing by appointment only. Contact the duty biochemist (6366) <u>at least a week in advance of lumbar puncture</u> to arrange for a member of staff to collect samples into liquid nitrogen.**	Sent away (London)	See report for ranges and interpretation of specific metabolite pattern.
Free Fatty acids and beta hydroxybutyrate, CPD code = BIONEFA Investigation of hypoglycaemia and fatty acid oxidation defects.	0.5 mL in **Fluoride Heparin**. Separate sample required. Ideally measured alongside a plasma glucose level.	Sent away (Sheffield)	Results are interpreted in light of glucose and free fatty acid concentrations.

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Galactosaemia screening test Investigation of galactosaemia (inborn error of metabolism) and prolonged jaundice.	0.1 mL **Lithium Heparin** Send separate sample.	Sent away (Leeds)	See report. Please note, recent transfusion invalidates results.
Galactose-1-Phosphate Confirmatory test, which is only available to patients who have a positive galactosemia screen.	2mL **Lithium Heparin,** deliver sample a.s.a.p. **Must be collected on Monday-Thursday ONLY.** Separate sample required.	Sent away (Bristol)	See report <0.60 umol/g Hb for known galactosemia.
α-galactosidase / Fabry's screen For the diagnosis of Fabry's disease. **Definitive diagnosis in females requires genetic testing. Consent should be obtained at the point of blood collection in any female patient.**	0.5 mL **EDTA** or **Lithium Heparin** or dry blood spot on guthrie card (leave 4h before packaging). A statement about consent for genetic testing should handwritten on request forms i.e. 'genetic consent obtained/not obtained.' **Send to lab as soon as possible.**	Sent away (Manchester)	6.3-47 pmol/punch/hr
α-1,4 glucosidase / Pompe Screen Investigation of Pompe's disease	5mL **EDTA** **Samples MUST arrive at the laboratory Monday-Thursday to avoid delays in transport over the weekend.** Separate sample required.	Sent away (Manchester)	3 – 20 μmol/g.h with acarbose
β-glucosidase / Gaucher Screen Investigation of Gaucher's disease		Sent away (Manchester)	1-5μmol/g.h in white cells
Guanidinoacetate/ creatine metabolites (for disorders of creatine biosynthesis) Investigation of suspected creatine synthesis disorders (autistic spectrum disorder, speech and language delay, movement disorder)	**Lithium heparin** tube with 1mL paired random urine in a plain container (deliver within 2h of collection).	Sent away (Leeds)	See individual report
Homocysteine (CPD code = BIOHOM) Diagnosis of classical homocystinuria.	0.5mL **EDTA** **Send to lab immediately (must be within 30min).** Separate sample required.	Sent away (Leeds)	<18 umol/L
Karyotyping* Investigation of suspected aneuploidy and disorders of sexual development.	1-2 mL in **Lithium Heparin** **Please include completed genetics form*.** Separate sample ESSENTIAL.	Sent away (NEY Genomics hub)	See individual report.
Lead, CPD code = BIOLEA Suspected toxicity e.g. in PICA, anaemia, neurological disorders	1mL in **EDTA** tube (a special tube is no longer required)	Sent away (Leeds)	<2.4 umol/L
Manganese (blood) Suspected deficiency in patients on long-term TPN (bone demineralisation, poor growth) or toxicity in patients receiving trace element supplements (tremor, motor regression)	**Contact the duty biochemist (x6366) to arrange** for specialist collection tube to be delivered (certified trace element free tube)	Sent away (Leeds)	See individual report

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Mercury (blood) Suspected overexposure	2mL whole blood in **EDTA.** Separate sample required.	Sent away (Guildford)	See report <30 nmol/L
Microarray* Investigation of learning or behavioural difficulties, delay, autism, seizures or dysmorphism.	1mL in **EDTA** **Please included completed genetics form*.** Separate sample ESSENTIAL.	Sent away (NEY Genomics Hub)	See individual report.
Mitochondrial Genes* Investigation of maternally inherited disorders presenting with myopathy, deafness, blindness, seizures and acidosis.	2-3 x 4.5mL in **EDTA** **Please include completed mitochondrial request form*.** Separate sample required.	Sent away (Newcastle)	See individual report.
Mucopolysaccharide screen **Organic Acids**	Plain random urine (sent to Leeds) – please specify if particular disorder suspected		
Phenobarbitone Investigation of suspected toxicity or non-compliance.	0.5 mL **serum.** Requires a separate, pre-dose sample to arrive at the laboratory within 2 hours of collection.	Sent away (Hull)	See individual report.
Phytanic acid Investigation of suspected peroxisomal disorders – this test is also included in the white cell enzymes panel.	5mL whole blood in **EDTA** **Samples MUST arrive at the laboratory Monday-Thursday to avoid delays in transport over the weekend.**	Sent away (Manchester)	See individual report.
Porphyrin (red cells, CPD code = BIOBPORQ). Increased levels seen in erythropoietic protoporphyria , congenital erythropoietic porphyria, iron deficiency and lead poisoning.	0.5mL in **EDTA WITH** Random Urine **AND** Faecal samples **Samples MUST be protected from light (e.g. brown envelope, wrap in tinfoil)** Send separate samples.	Sent away (Cardiff)	Full interpretation of plasma porphyrin profile provided on report
Selenium ***Included in Trace Elements Profile with copper and zinc.*** For assessment of nutritional deficits.	0.5mL **Serum** Separate sample required. No add-ons permitted.	Sent away (Leeds)	<6 months: 0.4-0.7 umol/L 7m-6y : 0.6-1.2 umol/L >6 years: 0.8-2.0 umol/L
Sweat Test* Used in the diagnosis of cystic fibrosis	**Testing by appointment only. Please contact the laboratory to arrange (see page 12)**	Twice weekly	Sweat Chloride (mmol/L) <30: CF unlikely 30-60: Indeterminate >60: Supports a diagnosis of CF
Trace Elements ***Includes copper, zinc and selenium,*** *CPD code = BIOSELE.* Measured in patients at high risk of nutritional deficiency e.g. parenteral nutrition or highly restricted diet.	1mL **Serum** Send separate sample. Not permitted as an add-on request due to risk of sample contamination.	Sent away (Leeds)	Selenium (umol/L): 0-6 months: 0.4 - 0.7 7m – 6 y: 0.6 - 1.2 >6 years: 0.8 - 2.0 Copper / Zinc: See report for age-dependent limits
Transferrin glycoforms (sialotransferrin) CPD code = BIOTRAG Investigation of congenital disorders of glycosylation (CDG),	1mL **Serum** Send separate sample	Sent away (London)	See report

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Very long chain fatty acids (VLCFA) including phytanic acid Investigation of suspected adrenoleukodystrophy and adrenomyeloneuropathy (ALD and AMN).	1mL **Lithium Heparin** Send separate sample.	Sent away (Sheffield)	C22: 15-112 umol/L C24: 14-80 umol/L C26: 0.33-1.50 umol/L C24/22 ratio: 0.44-0.97 C26/22 ratio: 0.005-0.030
Vitamin A Measured as part of a nutritional screen in patients at risk of deficiencies of fat soluble vitamins (e.g. in cystic fibrosis)	2mL **Serum** or **Lithium Heparin** Send separate sample. **Please protect from light if there is a delay in sending to the laboratory e.g. wrap securely in tinfoil or place in a brown paper envelope.**	Sent away (Hull)	1.1-2.6 umol/L (adult) 0.7-1.7 umol/L (<12y)
Vitamin E As for vitamin A, above			12-42 umol/L (adult) 7 – 21 umol/L (<12y)
White Cell Enzymes, CPD code = BIOWCE A screen of enzyme activities which are associated with 17 different lysosomal storage diseases.	5mL **EDTA** **Samples MUST arrive at the laboratory Monday-Thursday to avoid delays in transport over the weekend.**	Sent away (Willink)	See individual enzymes. If 5mL blood unavailable, please state any enzymes / disorders which are a priority.

*** Further information on sweat testing, CSF Neurotransmitter collection and Genetic testing can be found on pages 12-13**

URINE TESTS (ROUTINE AND SPECIALIST)

The table below lists all tests performed in urine, including specialist tests for oncology, endocrinology, neurology, urology and metabolic medicine. For renal stone analysis, see 'calculus' under Specialist and Metabolic Investigations').

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
AASA – alpha amino adipic semi aldehyde Investigation of pyridoxine-responsive seizures.	5mL random urine – hand deliver immediately and mark as URGENT, FAO Duty Biochemist	Sent Away (London)	See report
Amino Acids - *Part of Metabolic Screen* Diagnosis of disorders of amino acid metabolism; urine is particularly important for diagnosis of cystinuria, which can be missed using plasma only.	1.5 mL random urine **Please state whether a specific disorder is suspected.**	Weekly	See report for specific pattern of results and interpretation.
Albumin:creatinine ratio (**ACR** or **urine microalbumin**) An indicator of glomerular damage in patients with protein-losing renal disease.	2mL random urine in plain container. Early morning "first pass" urine is preferred.	Daily	<30 mg/mmol creatinine (non-diabetic)
Calcium:creatinine ratio, CPD code = BIOUCALR Investigation of hyper- or hypo-calcaemia, where a congenital renal cause is suspected (e.g. familial hypocalciuric hypercalcaemia). Investigation of renal stone formation.	2mL random urine in a plain container.	Daily	See report (age related)
Catecholamines, CPD code = BIOUCAT Diagnosis / monitoring Neuroblastoma only – **if investigating early onset hypertension, please request urine metanephrines** / BIOUMET (catecholamines unsuitable).	2mL random urine in acidified collection container (packs located on CAU, ward 17, SGH Rainbow ward and Bridlington outpatients or available from pathology reception). **Please provide relevant clinical details.**	Sent away (Leeds)	See report (age related). HMMA and VMA ratio to creatinine measured to account for differences in urine concentration.
Chloride (urine) Only measured in some circumstances - contact laboratory if unsure.	2mL random urine in plain container or plain 24hr urine collection.	Daily	170-250 mmol/L OR 170-250 mmol/24hr
Copper (urine) Increased in Wilson's disease	24h urine	Sent away (Guildford)	<0.9 umol/24h
Citrate (urine), CPD code = BIOUCITR Investigation of renal tubular acidosis and renal stone disease.	2mL random urine in a plain container.	Weekly	0.11-1.75 mmol/mmol creatinine
Cortisol (urine free) Useful in the diagnosis of Cushing's syndrome	Plain 24h urine collection. **Please list any medications.**	Weekly	0-165 nmol/24h

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Creatinine (urine) Used as a measure of renal function (creatinine clearance) and to correct for differences in urine concentration when making measurements in random urine.	2mL random urine in plain container or plain 24hr urine collection.	Daily	F = 8-13 mmol/24h M = 8-16 mmol/24h N.B. Age and muscle mass dependent.
Creatinine clearance An approximate measurement of glomerular filtration rate	Plain 24h urine collection **AND** a **serum** sample drawn on the same day	Daily	Age related, see report
Cystine Renal stones, suspected cystinuria	2mL random urine. **Deliver to the lab immediately.**	Sent away (Leeds)	Child: <8 umol/mmol creat Adult: <15 umol/mmol creat
Drugs of Abuse Screen Investigation of passive or active drug exposure e.g. confirmation of opiate or methadone exposure in neonates with withdrawal symptoms, suspected drug ingestion in children or adolescents.	5mL random urine **This is NOT a forensic service – use should be strictly confined to situations where drug detection will affect clinical management.**	Daily	Results given in positive/negative format. **Please contact the duty biochemist (6366) if testing for a specific drug or if sample storage is required for forensic purposes.**
Guanidinoacetate (urine) – Screen for Creatine Synthesis Disorders, CPD code = BIOCSD Investigation of suspected creatine biosynthesis disroders in children with seizures, intellectual disability and speech delay.	2mL random urine in a plain container & **paired 1mL blood sample** in **lithium heparin** tube. **Deliver to lab within 2h of collection.**	Sent away (Leeds)	See report for interpretation of metabolite patterns in blood and urine. Urine is essential for diagnosis of the commonest (X-linked) disorder, plasma rules out rarer forms.
Magnesium (urine, CPD code = BIOUMAGR) Estimation of renal magnesium loss in deficiency states	2mL random urine in a plain container.	Daily	See report (age related)
Mercury (urine) In suspected toxicity or exposure to inorganic mercury compounds	24h urine	Sent away (Guildford)	<50 nmol/24 hr
Metabolic Screen ***Includes urine amino acids, urine organic acids and urine sugar chromatography.***	5mL random urine in a plain container. If glycosaminoglycans are required, please state.	Sent away (Leeds)	See individual analytes
Metanephrines (urine)	24h urine (preferred) or 5mL random urine Investigation of paediatric hypertension or adrenal incidentaloma	Sent away (24h to Hull or random urine to Leeds)	See individual reports
Mucopolysaccharide screen One of the components of the metabolic screen test. Increased levels are seen in patients with mucopolysaccharidoses.	3mL random urine in a plain container.	Sent away (Leeds)	See report – pattern of results may be normal or suggestive of a specific disease.

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Organic acids (urine) - *Part of Metabolic Screen* Detection of a wide variety of metabolic disorders.	5mL random urine in a plain container. **Please state whether a specific disorder is suspected and list any medications.**	Sent away (Leeds)	See report - pattern of results may be normal or suggestive of a specific disease.
Orotic Acid (urine) Suspected urea cycle disorders; X-linked ornithine transcarbomylase deficiency cannot be diagnosed using plasma or urine amino acids.	2mL random urine in a plain container.	Sent away (Leeds)	See report - pattern of results may be normal or suggestive of a specific disease.
Osmolality (urine) Investigation of polyuria	2mL random urine in a plain container.	Daily	300-900 mmol/kg
Oxalate (CPD code = BIOUOXAR) Investigation of renal stones	2mL random urine in a plain container. **Deliver to the lab immediately.**	Weekly	See report or for age-related ranges
Phosphate (urine) Investigation of abnormal calcium / phosphate levels or renal stones.	2mL random urine in a plain container.	Daily	See report or for age-related ranges
Porphobilinogen (urine, CPD code = BIOUPBGCR) Screening test for acute porphyria.	2mL fresh random urine in a plain container – **must be protected from light and collected during an attack if possible**. **Please contact the duty biochemist to discuss all requests** and list relevant medication and history.	Sent away (Cardiff)	See report for full details. N.B normal levels may be observed between acute attacks.
Porphyrin (urine, CPD code = BIOUPOR) Quantitative test for more detailed investigation of porphyria. A full screen requires urine, stool and blood samples.	5mL fresh random urine in a plain container - **must be protected from light.** List medications and clinical / family history.	Sent away (Cardiff)	See report for full interpretation of metabolite pattern.
Potassium (urine) Estimation of potassium losses in potassium deficiency states	2mL random urine in a plain container **AND** a **serum** sample for U&E	Daily	30-125 mmol/L Interpret in light of serum potassium results.
Protein (urine) Estimate of renal protein losses in patients with renal failure and nephrotic syndrome	2mL random urine in a plain container or plain 24h urine collection.	Daily	As a guide only: <20 mg/mmol creatinine
Sodium (urine) Measure of sodium excretion in patients with electrolyte disturbances	2mL random urine in a plain container or plain 24h urine collection.	Daily	40-300 mmol/L OR 50-250 mmol/24 hr Interpret in light of serum sodium results.
Sugar Chromatography (urine) - *Part of Metabolic Screen* Investigating disorders of carbohydrate metabolism	2mL random urine in a plain container. Please provide clinical details.	Sent away (Leeds)	See report for full interpretation of metabolite pattern.

ANALYTE	SAMPLE REQUIREMENTS	ANALYSED	REFERENCE RANGE
Steroid Profile (urine) Diagnosis and monitoring of congenital adrenal hyperplasia and other disorders of steroid metabolism	2mL random urine in a plain container OR plain 24h urine collection. Provide full clinical and medication details.	Sent away (London)	See report for full interpretation of metabolite pattern.
Stone screen Investigation of recurrent renal stone formation	5mL random urine in a plain container. **Deliver to the lab immediately.**	Screen includes calcium, phosphate, magnesium, citrate, oxalate, urate – see individual entries for details.	
Urate (urine) Measured in 24h urine as part of screen for renal stone disease.	2mL random urine in a plain container.	Daily	See full report (age related)

FAECAL (STOOL) TESTS

ANALYTE (TEST SET)	SAMPLE REQUIREMENTS	FREQUENCY OF ANALYSIS	REFERENCE RANGE
Calprotectin (faeces, CPD code = BIOFCAL) Marker of inflammatory bowel disease	Fresh random faecal sample Samples should ideally be the size of a grape or small marble. Note that liquid stool samples are unsuitable for analysis, and blood staining may affect results.	Twice weekly	Poorly defined – levels >100 ug/g usually warrant further investigation
Faecal elastase, CPD code = BIOFELA - Measure of exocrine pancreatic function		Weekly	>200 µg/g faeces
Porphyrin (faeces, CPd code = BIOFPORQ) Investigation of suspected porphyria, particularly if cutaneous symptoms present.	Fresh random faecal sample (10g - 15g) – **must be protected from light. Please provide clinical details.**	Sent away (Cardiff)	See report for full details – specific pattern of metabolites may be normal or suggestive of a specific disease.

Faecal Reducing Substances are no longer tested at the laboratory. Due to problems with sample stability and the sensitivity and specificity of traditional test methods, an exclusion diet is now considered a better means of diagnosing lactose intolerance and other malabsorbtive conditions.

4. INVESTIGATIONS BY CONDITION

The following sections are aimed at helping the local laboratory and clinical teams carry out a concise but comprehensive panel of investigations for specific presentations of suspected metabolic origin. Where possible, clear causes or causes based on clinical symptoms or history alone should be ruled out prior to testing. Protocols are Best Practice Guidelines formulated by an expert opinion group at the Metabolic Biochemistry Network (MetBioNet).
Further information is available via the Metabolic Biochemistry Network website:

https://metbio.net/best-practice-guidelines/

HYPOGLYCAEMIA

Instructions and sampling kits for the investigation of hypoglycaemia in children are available in the following locations:

YORK HOSPITAL	SCARBOROUGH HOSPITAL
SCBU	SCBU
Ward 17	Rainbow Ward
Child Assessment Unit	Paediatric Outpatients
ED Resus	ED Resus

Kits are housed in a labelled, grey, hard plastic box bearing a blue Roche logo:

If you require a further kit, please contact the Point of Care team on:

YORK HOSPITAL: ext. 5890 (9am-5pm)

SCARBOROUGH HOSPITAL: ext. 2659 (9am-5pm)

A set of biochemistry tests is available to order as a panel in CPD **(Hypoglycaemia screen – PA).** The Hypoglycaemia screen in CPD requires the following samples:

Tests included in panel	Samples required
AMINO ACIDS	One full **Serum** or **Lithium Heparin** tube
CALCIUM - ADJUSTED FOR ALBUMIN , LFT (LIVER FUNCTION TESTS) U&E INCLUDING EGFR	One full **Serum** or **Lithium Heparin** tube should be sufficient for UE, LFT and Calcium
ACYL CARNITINE and CARNITINE	Dried blood spot on guthrie card
AMMONIA	**EDTA** (hand deliver to lab within 30 minutes of collection, up to 60 minutes for samples on ice)
CORTISOL	**Serum** or **Lithium Heparin**
C PEPTIDE AND INSULIN	**Serum** tube (hand deliver to lab within 1 hour of collection)
GROWTH HORMONE	**Serum** or **Lithium Heparin**
LACTATE GLUCOSE	**Fluoride Heparin** - One full tube should be sufficient for Glucose and Lactate
FREE FATTY ACIDS / BETA HYDROXYBUTYRATE	One full **Fluoride Heparin** tube
AMINO ACIDS – URINE, ORGANIC ACID SCREEN – URINE	A minimum of 5mL random urine collected into a plain universal container (referred to Leeds)

https://metbio.net/wp-content/uploads/MetBio-Guideline-FUJA773994-19-11-2018.pdf

SEIZURE DISORDERS

The following tests are available to order on CPD using the '**Seizure-PA'** panel:

Tests included in panel	Samples required
BONE PROFILE	2 x full **Serum** or **Lithium Heparin** tubes should be sufficient to carry out all of the necessary tests
CRP (C-REACTIVE PROTEIN) - SERUM	
GLUCOSE - SERUM (BROWN TUBE)	
MAGNESIUM - SERUM	
U&E INCLUDING EGFR - SERUM	

The guideline below provides further information on the metabolic investigations which should be performed in children with ongoing seizure disorders

https://metbio.net/wp-content/uploads/MetBio-Guideline-DUHA851763-19-11-2018.pdf

HYPERAMMONAEMIA

The following guideline provides a framework for the metabolic investigation of neonates or children presenting with unexplained hyperammonaemia

https://metbio.net/wp-content/uploads/MetBio-Guideline-PERE918546-10-12-2018.pdf

CARDIOMYOPATHY

The following guideline provides a framework for the metabolic investigation of neonates or children presenting with unexplained cardiomyopathy

https://metbio.net/wp-content/uploads/MetBio-Guideline-NAFF997813-13-07-2012.pdf

PROLONGED JAUNDICE

The following panel is available to order on CPD named '**Prolonged jaundice -PA**':

Tests included in panel	Samples required
LFT (LIVER FUNCTION TESTS) - SERUM	**Serum** or **Lithium Heparin** tube
FULL BLOOD COUNT (PAEDIATRIC)	**EDTA** tube
G-6-PD SCREENING TEST	Contact haematology for details if required

The following guideline provides a framework for the metabolic investigation of neonates or children presenting with unexplained prolonged jaundice.

https://metbio.net/wp-content/uploads/MetBio-Guideline-DUUM428158-28-01-2013.pdf

GLOBAL DEVELOPMENTAL DELAY

A comprehensive set of tests for the investigation of developmental delay is available as an order set CPD (**Developmental Delay – PA**). This requires the following samples:

Tests included in panel	Samples required
AMINO ACIDS - SERUM	**Serum** or **Lithium Heparin** (referred to Leeds)
CALCIUM - ADJUSTED FOR ALBUMIN, CREATINE KINASE, FERRITIN LFT (LIVER FUNCTION TESTS), GAMMA GLUTAMYL TRANSFERASE (GGT), U&E INCLUDING EGFR URATE	2 x full **Serum** or **Lithium Heparin** tubes should be sufficient to carry out all of these tests
LEAD - BLOOD	Separate **EDTA** tube (referred to Leeds)
THYROID FUNCTION TEST (TSH AND FT4) FERRITIN	1 x full **Serum** or **Lithium Heparin** tube should be sufficient
AMINO ACIDS – URINE, MUCOPOLYSACCHARIDE SCREEN – URINE ORGANIC ACID SCREEN – URINE, SUGAR CHROMATOGRAPHY - URINE CREATININE – URINE	A minimum of 5mL random urine collected into a plain universal container (referred to Leeds)
CHROMOSOME STUDIES - ST JAMES	Separate **lithium heparin** tube and completed genetics request form (see **page 13**) - referred to Leeds
FULL BLOOD COUNT (PAEDIATRIC)	Separate **EDTA** tube
DNA STUDIES (if not already performed, check CPD for paperwork)	Separate **EDTA** tube and completed genetics request form (see **page 13**) - referred to Leeds

The following tests are **NOT** part of the CPD panel but **should always be considered**:

Tests	Inclusion criteria	Samples required
Biotinidase	Patients with elevated lactate levels or clinical symptoms such as increased work of breathing / persistent wheeze, conjunctivitis seizures, hair loss, visual or hearing defects, skin rash	0.2 mL in **Serum** **Samples must be delivered to laboratory immediately.**
Total Homocysteine	ANY patient with symptoms suggestive of homocystinuria i.e. myopia, lens abnormalities, abnormally long bones, FH of cardiovascular disease Patients born outside the UK or within the UK BEFORE Jan 2015 (i.e. those who have NOT undergone expanded newborn screening).	Minimum of 0.5 mL plasma in an **EDTA** tube – to be delivered to the lab immediately and arrive within 30 min of collection. Do not use the pod system.
Guanidinoacetate / creatine metabolites	Patients from families in which 2 or more males are affected by global developmental delay ANY patient with developmental delay and significant speech and language delay	**Lithium heparin** tube with 1mL paired random urine in a plain container (deliver within 2h of collection).

https://metbio.net/wp-content/uploads/MetBio-Guideline-GEPE598185-30-11-2020.pdf

FACIAL DYSMORPHISM

The following guideline provides a framework for the metabolic investigation of neonates or children presenting with unexplained dysmorphic features

https://metbio.net/wp-content/uploads/MetBio-Guideline-PEDE620597-28-01-2013.pdf

SUDDEN UNEXPECTED DEATH IN INFANCY (SUDI)

Investigation of unexpected infant deaths requires careful collection and handling of a diverse range of sample types. To facilitate this process, **'SUDI boxes' are available** in the following locations:

YORK HOSPITAL	SCARBOROUGH HOSPITAL
ED resus room	ED paediatric resus room
Mortuary	Rainbow treatment room

These contain sample containers for collection of blood, urine/nappy and nasal swabs as well as a document for completion by the clinician dealing with the death. Completed samples and paperwork should be placed back into the SUDI box, sealed with a tamper-proof tag and sent urgently to the Laboratory Medicine for processing.

Please notify the duty biochemist on 1904 72 6366 (or the laboratory if calling out of hours) if a SUDI box is to be delivered to the laboratory. You will be asked to sign a

chain of custody form on delivering the box, and should obtain a new, unused box in it's place.

For further information on metabolic causes of SUDI, please refer to:

https://metbio.net/wp-content/uploads/MetBio-Guideline-RASU337946-27-11-2010..pdf