<table>
<thead>
<tr>
<th>CONTENTS</th>
<th>Page Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Address, telephone numbers and enquiries</td>
<td>3</td>
</tr>
<tr>
<td>Services provided</td>
<td>4</td>
</tr>
<tr>
<td>Laboratory opening times</td>
<td>4</td>
</tr>
<tr>
<td>Requests for analyses</td>
<td>5</td>
</tr>
<tr>
<td>Urgent requests</td>
<td>7</td>
</tr>
<tr>
<td>Specimen transport</td>
<td>7</td>
</tr>
<tr>
<td>Quality Assurance</td>
<td>10</td>
</tr>
<tr>
<td>Specialised services</td>
<td>11</td>
</tr>
<tr>
<td>Inborn errors of metabolism</td>
<td>11</td>
</tr>
<tr>
<td>Molecular Genetics</td>
<td>13</td>
</tr>
<tr>
<td>Pre-natal diagnosis</td>
<td>14</td>
</tr>
<tr>
<td>Newborn Screening</td>
<td>14</td>
</tr>
<tr>
<td>Table of Analyses</td>
<td>16</td>
</tr>
<tr>
<td>Turnaround times</td>
<td>17</td>
</tr>
<tr>
<td><strong>A-Z Tests</strong></td>
<td>18</td>
</tr>
<tr>
<td>Details of charges for metabolic tests</td>
<td>29</td>
</tr>
</tbody>
</table>
Clinical Chemistry

**Telephone numbers & enquiries**

Hospital switchboard: 0114 271 7000
Laboratory Fax: 0114 270 6121

Direct Telephone

Dr J R Bonham, Consultant Clinical Scientist & HoD
Clinical Chemistry 2717404
Dr Bonham’s Secretary – Lynne Darwin 2717318
Mr D A Wardley, Laboratory Services Manager 2717444
Laboratory Secretary – Alison Lenthall 2717340

Duty Clinical Scientist Bleep No 095
(From outside the hospital please dial the switchboard and request bleep 095)

**Metabolic Section**

Result enquiries 2717445
Dr Simon Olpin (Metabolic Lead) 2717267
Camilla Scott (Clinical Scientist) 2717307
Nigel Manning (Clinical Scientist) 2717479/2717307
Joanne Croft (Clinical Scientist) 2717307
Dr Jane Dalley (Clinical Scientist) 2260972/2717479/2717307
Jenny Watkinson (Lead Biomedical Scientist) 2717445
Louisa Edwards (Biomedical Scientist) 2717405

**Tissue Culture Section**

Dr Simon Olpin (+ answer phone) 2717267
Prenatal diagnosis enquiries 2717267

**Newborn Screening Section**

Melanie Downing (Screening Lead Scientist) 2717302
Catherine Dibden (Clinical Scientist) 2717346
Lynette Shakespeare (Clinical Scientist) 2717346
Joyce Baston (Lead Biomedical Scientist) 2717500
Sheila Ellin (Biomedical Scientist) 2717346
Martin Maloney (Biomedical Scientist) 2717346
Newborn Screening Results (08:30-12:30) 2717257
Answering Machine and Fax 2717263
Sheffield Diagnostic Genetics Service

Telephone numbers & enquiries
Ann Dalton (Head of Laboratory)  2717004
Richard Kirk (Lead Clinical Scientist – Inborn Errors of Metabolism)  2267023
Liz Allen (Clinical Scientist – Inborn Errors of Metabolism)  2717003
Jo Martindale (Clinical Scientist – Neurodegenerative/Mitochondrial Diseases)  2260723
Laboratory fax  2750629
Email  SDGS@sch.nhs.uk

SERVICES PROVIDED

A Specialised service is provided as follows:

Inborn Errors of Metabolism

A regional service is provided by Clinical Chemistry for the investigation of suspected metabolic disorder. This service is available to the Sheffield Children’s NHS Foundation Trust without cross charging and to other users on a cost per test basis (see pages 29-32).

Newborn Screening

Screening covers all babies born in the East Midlands SHA, South Yorkshire and South Humberside portion of the Yorkshire and Humber SHA (Derbyshire, Leicestershire, Lincolnshire, Northamptonshire, Nottinghamshire, Rutland, and South Yorkshire). They are tested for phenylketonuria (phenylalanine), congenital hypothyroidism (TSH), cystic fibrosis (immunoreactive trypsin), medium chain acyl CoA dehydrogenase (octanoylcarnitine) and sickle cell disorders (haemoglobin profile) using dried blood spots from the newborn screening cards.

Sheffield Diagnostic Genetics Service

Molecular Genetics is the analysis of the detailed structure of the Human genome. The aim is to establish the mutation or mutations that have given rise to the disorder in that individual or family. The laboratory investigates both familial and sporadic conditions, including molecular changes that give rise to malignant conditions.

NORMAL LABORATORY OPENING TIMES

Clinical Chemistry, Newborn Screening  Monday to Friday
9:00am- 5:00pm
Sheffield Diagnostic Genetics Service  Monday to Friday
9:00am- 5:30pm
REQUESTS FOR ANALYSES

Legible request forms (see below) must accompany all samples. Every sample for which an analysis is required, other than those for routine newborn screening, must be accompanied by a FULLY COMPLETED laboratory request form signed by the doctor making the request and giving his/her bleep number. It is also important to include the time and date on which the sample was collected plus clinical details.

Minimum Request/Referral Form Labelling Criteria:-

- *NHS number or A/E number/ Hospital registration number
- Patient’s full name including first name (family name in capitals)
- Date of birth
- Location where results are to be sent
- Consultant (or referring laboratory)
- Test required
- Sample type
- *Important identifier

Highly desirable information includes:-

- Date and time of collection (particularly if more than one sample is likely to be obtained on the same day)
- Clinical details (including provisional diagnosis and current drug therapy)
- Clinician’s bleep number
- Patient’s address including postcode
- Patient’s gender

For urgent or telephoned requests, it is helpful to have the signature of medical officer, and the legible printed name for urgent or telephoned results

LABELLING OF PATHOLOGICAL SAMPLES

Sample and request form information must be compatible
Samples will only be accepted if minimum criteria are met. This responsibility lies with the person collecting the sample. Failure to meet these requirements may result in the sample being rejected.

As defined by laboratory policy all pathological samples sent to the laboratory must contain a minimum of the following information:

Minimum Criteria

Primary sample i.e. container the sample was originally collected into
Must contain the following identifiers

1. Surname/family name
2. Forename (or Baby, Twin 1 etc, if forenames have not been given)
3 At least one of the following
• *NHS number or A/E number
• Date of birth
• Hospital registration number
*Important identifier

Samples for monitoring must also have
• Date sample taken
• Sample type

Sub sample e.g. plasma or serum

Must contain at least two of the following identifiers:
• *NHS number
• A/E number
• Hospital registration number
• Surname/family name
• Forename (or Baby, Twin 1 etc, if forenames have not been given)
• Date of birth
• Requesting laboratory number
*Important identifier

SPECIMEN CONTAINERS

Please can we also draw your attention to requirements for specimen containers. We have in the past, accepted samples in containers that were unsuitable for our sample storage system and have transferred samples into our own containers. This is a potential risk. We would therefore ask you to strictly comply with the size of containers that we now stipulate for our investigations. We also request the discontinuation of the use of “push-on” tops for samples sent to us, as this introduces extra risk for sample contamination to our laboratory staff and leads to the loss of sample volume. For plasma samples the maximum height of the container should not exceed 65 mm. For urine containers the height should not exceed 100 mm and width should not exceed 30 mm.

Legal Responsibilities

In signing a request form the person making the request assumes responsibility under Section 7 of the Health and Safety at Work Act and will be assumed to be familiar with its requirements in relation to danger of infection.

To fulfil these regulations you must comply with the following:

1 All samples must be in a sealed plastic bag and the request form placed in the separate compartment provided.
2 Full and appropriate clinical details and danger of infection labels on both request form and sample are required from Category 3 risk patients.
3 Samples from patients with suspected and proven HIV infection must also be enclosed in a cardboard box.
4 Data on request form may be stored on laboratory computer files. It is assumed the person completing the form has done so in accordance with the requirements of the Data Protection Act 1984.

The attention of medical staff is drawn to the warning notice printed upon each laboratory request form concerning specimens which might carry a risk of infection. The doctor completing the request should also indicate on the form if the patient has a communicable disease such as rubella, for the protection of any laboratory staff who might attend the patient.

**URGENT REQUESTS**

Urgent requests must be arranged with the laboratory by telephone so that if there is any delay in receipt, steps can be taken to locate the sample. Urgent samples which arrive in the laboratory without prior arrangement run the risk of being delayed, as they will be analysed routinely.

Some tests may be performed out of hours after reference to the Consultant Clinical Scientist.

**SPECIMEN TRANSPORT**

Specimens must be sent to the laboratory contained in a transparent leak proof plastic bag. The request form must be separated from the specimen. Any label indicating a danger of infection must be shown on the request form.

Urgent samples must be arranged with the laboratory before dispatch and *sent by courier*

Non-urgent samples

Suitable postal or other delivery arrangements must be made by the sending laboratory. Samples must be sent direct to the laboratory; we cannot undertake to collect samples from rail stations or other collection points. Contact the laboratory before dispatch if the request is unusual or urgent. Details of sample preservation and packaging are given below. Charges are detailed on pages 29-32.

Post Office regulations require that all pathological samples are sent by first class post. The use of second class letter or parcel post is specifically forbidden. Padded envelopes used alone without a suitable inner container are not permitted. The regulations (RML 12/87) are summarised below.

1 Hazard group 4 pathogens are prohibited, other pathological specimens may be sent provided that they comply with the regulations.
2 Specimens may be sent by qualified medical, dental or veterinary practitioners, a registered nurse, a recognised laboratory or institution.
Members of the public may not send such specimens unless requested to do so by one of the above who must supply them with the required packaging and instructions.

Only first class letter or Datapost may be used.

There is a range of acceptable packaging but the following must be observed.

Every specimen must be in a primary container hermetically sealed or otherwise securely closed. The capacity of the primary container must not exceed 50 mL unless specifically permitted. The primary container must be wrapped in enough absorbent material to absorb all possible leakage, and sealed in a leakproof plastic bag.

The container and its immediate packaging must be placed in one of the following.

a) a polypropylene clip-down container
b) a cylindrical light-metal container
c) a strong cardboard box with a full-depth lid
d) the appropriate groove in a two piece polystyrene box, empty spaces must be filled with absorbent material, the box must be secured with self-adhesive tape.

A padded outer bag is recommended.

Soft absorbent packaging must be used between samples to prevent contact.

Written agreement from the Post Office is required for non-standard packaging.

The outer packaging must be labelled ‘PATHOLOGICAL SPECIMEN - FRAGILE WITH CARE’ with the name and address of sender.

Therapeutic and diagnostic materials such as blood products are accepted under the same conditions.

Packets found in the post which contravene the regulations will be detained and may be destroyed. Any person who sends deleterious substances without conforming to the regulations may be liable to prosecution.

Please note. Infectious pathology samples may only be transported in packaging which meets the U.N. class 6.2 specifications and the 602 packaging requirements. These new packaging requirements are described below:
BASIC TRIPLE PACKAGING SYSTEM.

The system consists of three layers as follows:

Primary receptacle

A labelled primary watertight, leak-proof receptacle containing the sample. The receptacle is wrapped in enough absorbent material to absorb all fluid in case of breakage.

Secondary receptacle

A second durable, watertight, leak-proof receptacle to enclose and protect the primary receptacle(s). Several wrapped primary receptacles may be placed in one secondary receptacle. Sufficient additional absorbent material must be used to cushion multiple primary receptacles.

Outer shipping package

The secondary receptacle is placed in an outer shipping package which protects it and its contents from outside influences such as physical damage and water while in transit.

Information concerning the sample, such as data forms, letters and other types of information that identify or describe the sample and the identity of the shipper and receiver should be taped to the outside of the secondary receptacle.

Newborn Screening (Guthrie Cards)

By common consent these regulations are deemed inappropriate for dried blood specimens on Newborn Screening (Guthrie) cards. The blood spots should be allowed to dry thoroughly before packing, the card placed in the transparent paper (Glassine) envelope provided (not plastic as this may cause the specimen to “sweat”) and sent by, first class post, in a stout envelope as if it were a normal letter or in a newborn screening pre paid envelope according to local arrangements.
QUALITY ASSURANCE

The Department participates in national external quality assurance schemes to monitor the accuracy and precision of its analyses. Internal quality control is used to check the validity of results on a day to day basis. The laboratory computer also checks the credibility of individual results.

*It is important that the laboratory be informed at once if results appear inconsistent with a patient’s condition or are at variance with previous results.*
SPECIALISED SERVICES

Investigation of Inborn Errors of Metabolism

A service is provided for the detection, diagnosis and monitoring of patients with inborn errors of metabolism. Analyses performed include:

- Acylcarnitine profile
- Amino acids
- Ammonia
- Bile Salts
- Biotinidase
- Carnitine
- Cholestanol
- 7-Dehydrocholesterol
- Dimethylglycine
- Free fatty acids
- Galactitol
- Galactosaemia Screen
- 2OH glutaric acid (chirality)
- Glutarate (quantitative)
- Glycerate (chirality)
- Glycosaminoglycans (screen and electrophoresis)
- Hexanoylglycine (quantitative)
- Homocysteine (total)
- Homocysteine (free)
- HVA/VMA (quantitative)
- 3-hydroxybutyrate
- 4-hydroxybutyrate (quantitative)
- Isovalerylglycine (quantitative)
- Lactate (including determination of chirality)
- Methyl citrate
- Methylmalonate (quantitative)
- Organic acids
- Orotic acid
- Phenylalanine
- Phytic acid
- Phytosterols
- Pipecolic Acid
- Plasmalogens (C₁₆ and C₁₈)
- Pristanic Acid
- Pyroglutamate (quantitative)
- Pyruvate
- Reducing substances
- Sweat test
- Thiosulphate
- Trimethylamine and oxide
- Very long chain fatty acids
Qualitative urine screening tests for glucose, reducing substances, cystine and homocystine, are also available.

It is important that requests for the investigation of inborn errors of metabolism are accompanied by adequate clinical information including drugs being taken at the time of sampling. If the relevant clinical information is detailed, the laboratory should be contacted by letter or telephone.

Further investigation of some disorders requires the use of cultured fibroblasts. The following are routinely available:

- Screen for disorders of long-or medium-chain fatty acid oxidation. This screen will detect defects of carnitine transport and deficiency of carnitine-palmitoyltransferase types 1 and 2, carnitine acylcarnitine translocase deficiency, very-long- or medium-chain acyl-CoA dehydrogenases, long-chain 3-hydroxyacyl-CoA dehydrogenase and other disorders of the trifunctional enzyme complex and mild to severe multiple acyl-CoA dehydrogenation defects (ethylmalonic-adipic aciduria and glutaric aciduria type 2).

* Carnitine-acylcarnitine translocase
* Glutaryl-CoA dehydrogenase (for glutaric aciduria type 1)
* Palmitoyl carnitine transferase Type I and II
* Propionyl-CoA carboxylase (for propionic acidemia)
* Pyruvate Carboxylase
* 3-Methylcrotonyl-CoA carboxylase
* Fumarate hydratase
* Release of $^{14}$CO$_2$ or $^{14}$C-incorporation from various substrates for the detection of isovaleric acidemia MSUD and other disorders
* Very long-chain fatty acids
* Citrulline incorporation into fibroblasts for detection of defects of argininosuccinate synthase and argininosuccinate lyase.
* Acylcarnitine profiles on cultured fibroblasts incubated with palmitate and L-carnitine.
* DHAP-AT activity for peroxisomal biogenesis disorders
* Filipin staining for Niemann Pick type C
* Catalase staining for peroxisomal biogenesis disorders.

Enquire for disorders not listed.

In general the laboratory will advise on the need for tissue based assays and make the necessary preliminary arrangements.
Sheffield Diagnostic Genetic Services

Details of all testing available are contained in the booklet “Investigative Protocols and Referral Guidelines” available from the Laboratory (Tel 0114 271 7003). Additionally, all services from the diagnostic laboratories, including Molecular Genetics are described on our website at http://www.sheffieldchildrens.nhs.uk/A-Z-List-of-Services.htm
Pre-natal diagnosis

Prenatal diagnosis may be performed in a variety of ways:

* Metabolite analysis on amniotic fluid (or occasionally chorionic villus or cultured fetal cells). We currently provide metabolite analyses for the diagnosis of methylmalonic aciduria, glutaric aciduria type II, isovaleric acidaemia, pyroglutamic aciduria (5-oxoprolinuria), Smith Lemli Opitz Syndrome, 4-hydroxybutyric aciduria, propionic aciduria and fumarate hydratase deficiency.

* Enzyme assay on chorionic villus (fresh or cultured) or cultured amniotic fluid cells. Most of the assays listed above for fibroblasts can be used with cultured amniotic fluid cells or chorionic villus material.

* DNA analysis

Prenatal diagnosis for other disorders is usually available in the UK but some conditions will require samples to be sent overseas.

Careful consideration of the technical aspects (timing, material and route, time of result and reliability) is an essential part of preparatory counselling and prenatal diagnosis should be arranged well in advance if possible. Reliable prenatal diagnoses requires that the initial diagnosis has been clearly established and it is important to appreciate the need for rigorous investigation even when the index case presents in a terminal phase with little hope of useful intervention.

Newborn Screening

Dried blood spot samples are collected between the 5th and 8th day of life by midwives to screen for phenylketonuria, congenital hypothyroidism, cystic fibrosis, sickle cell disorders and medium chain acyl CoA dehydrogenase deficiency. Results are sent out to the appropriate Child Health Records Department for entry into the Child Health Records Computer and checking against birth lists. Positive cases are referred for further investigation and treatment directly through their Family Practitioner to designated disorder specific clinicians. Individual negative results are NOT normally sent out to hospital doctors or Family Practitioners. If you wish to have a result returned to the ward or a particular doctor then the Newborn Screening dried blood spot specimen must be accompanied by a standard laboratory request form.

This service is largely separate from the routine analytical services offered in the hospital and in general it is NOT appropriate to enquire directly of the Newborn Screening Laboratory for a test result. If an abnormal result has been found then, as soon as it has been confirmed, the patient’s Family Practitioner and
designated clinician for that disorder will have been informed. If you have clinical suspicion of hypothyroidism it is better to initiate your own investigations since the neonatal test is only a screening assay and in any case will not detect secondary hypothyroidism. Similarly, suspicion of MCAD deficiency should always be vigorously pursued in the newborn period and separate investigations (including urinary organic acid analysis and acyl carnitine profile) are indicated. Immunoreactive trypsin is not always abnormal in cystic fibrosis patients with meconium ileus.
TABLES OF ANALYSES

Analyses are listed alphabetically. If the required analysis is not listed, check for pseudonyms. If the request is unusual, consult the laboratory. This section begins on page 18.

Units

Most results are reported in molar SI units but some drugs, hormones and proteins may be quoted in other units. Assistance with the conversion of results into alternative units can be obtained from the Duty Clinical Scientist (Bleep No 095).

Reference (normal) ranges

Reference ranges are provided for guidance in clinical decision making, rather than for prescriptive use. They are conventionally set to give the range of values which would be found in approximately 95% of a ‘normal’ population. They are derived from results obtained by this Department and from other sources. Reference ranges for blood refer to serum or plasma samples unless stated otherwise.

Changes during growth and development create age-related reference ranges for most analytes. Detailed ranges are kept in the Department and information upon them may be obtained from one of the Duty Clinical Scientists.

For the day to day interpretation of results age-related reference ranges have been condensed to cover generally recognised stages of development. These are generally printed automatically by the laboratory computer when the result is generated.

Newborn: First 7 days of life for term baby.

Neonate: First month of life for a term baby. Ranges may not apply to pre-term or small-for-dates babies.

Infant: Normally from the second month to one year, neonates are included in these ranges if not separately quoted.

Child: Normally one year to adolescence, neonates and infants are included in these ranges if not separately quoted.

Adult: From the end of adolescence

Accuracy and Imprecision of Results (Clinical Chemistry)

These are monitored and controlled by our quality assurance procedures. When patients are being repeatedly tested the significance of any apparent change in their results depends upon many factors including biological variability
(intrapersonal variation) and the imprecision with which an analysis is performed (analytical variation). To aid in the interpretation of consecutive results imprecision data can be generated from laboratory quality control data, which can then be used to determine whether two results are significantly different, or within the bounds of analytical variation. As a first approximation a result has a 95% probability (using a level of p< 0.05) of being genuinely different from a previous result for the same patient if the results differ by more than the quoted imprecision (2.8 times the analytical standard deviation).

For example:
Plasma urea result on day 1 = 6.0 mmol/L
Plasma urea result on day 2 = 6.8 mmol/L
Difference (day 2- day 1) = 0.8 mmol/L
Imprecision estimate for Urea = 0.84 mmol/L

Because the difference between the two results is less than the imprecision estimate there is a less than 5% chance that these results are statistically different. Of course, even if there is a significant analytical change between consecutive results this may well be within expected biological variation and consequently have little significance for the patient. It should be noted that analytical imprecision is not constant over the reportable range. Should you wish to discuss the significance of results further please contact the Clinical Scientist (Bleep 095).

**TURNAROUND TIMES**

<table>
<thead>
<tr>
<th>A</th>
<th>B</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ammonia</td>
<td>Acylcarnitine profile</td>
</tr>
<tr>
<td>Caffeine</td>
<td>Amino acid quantitation</td>
</tr>
<tr>
<td>Free fatty acids</td>
<td>Biotinidase</td>
</tr>
<tr>
<td>GIPUT screen</td>
<td>Carnitine</td>
</tr>
<tr>
<td>3-Hydroxy butyrate</td>
<td>HVA/VMA</td>
</tr>
<tr>
<td>Intermediary metabolites</td>
<td>Methylmalonate</td>
</tr>
<tr>
<td>Lactate</td>
<td>Mucopolysaccharides</td>
</tr>
<tr>
<td>Methotrexate</td>
<td>Organic acid</td>
</tr>
<tr>
<td>pH and reducing substances</td>
<td>Orotic acid</td>
</tr>
<tr>
<td>Pyruvate</td>
<td>Sweat test</td>
</tr>
<tr>
<td></td>
<td>Total homocysteine</td>
</tr>
<tr>
<td><strong>Turn around time 1 week</strong></td>
<td><strong>Turn around times 5-14 days</strong></td>
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<tr>
<td><em>(providing samples do not require repeat analysis)</em></td>
<td><em>(But if requested urgently, these analyses can often be performed more quickly)</em></td>
</tr>
</tbody>
</table>

**Turn around times for all other tests 4-6 weeks**

Occasionally TAT’s may be delayed following a bank holiday or other exceptional circumstances. However, if the analysis is urgent please phone the laboratory and we will do our best to obtain a timely result.
Acylcarnitine profile
Dried blood spot (Guthrie card) - at least two full circles and/or lithium heparin plasma NOT EDTA. However please note that for most situations a plasma sample for acylcarnitine is likely to be more informative. In addition to interpretive comments regarding the profile we give a quantitative result for free carnitine with each sample. (The newborn screening card for babies should already be in our possession - please contact the laboratory) if a new dried blood spot is being taken please ensure it is sent with a completed request form and sent to Clinical Chemistry not Newborn Screening. Send first class post Monday-Thursday.

Amino acids (screen)
urine: 10 mL aliquot of a random or 24 hour urine in a plain bottle; qualitative report given Store sample at -20°C. Send by first class post Monday-Thursday with normal packaging. Include information on current therapy.

Amino acids (quantitative)
plasma: µmol/L; Collect 1 mL blood into a lithium heparin tube, centrifuge as soon as possible (preferably at 4°C) separate plasma taking care not to disturb the buffy coat and store at -20°C. Age related reference ranges given with report.
urine: µmol/mmol creatinine; Random or timed collection. Store at -20°C, send 10 mL aliquot; reference ranges given with report Send by first class post Monday-Thursday with normal packaging. Include information on current therapy.

Amino acids in dried blood spot
Certain amino acids can be measured in dried blood spots for:
a) follow-up results from newborn screening programmes.
b) monitoring of selected patients receiving dietary treatment for inherited metabolic diseases. Contact the laboratory to arrange this. Dried blood spot (Guthrie card) - at least two full circles. This can be sent by first class post.
Amino acids in CSF
Serine and Glycine, Threonine, Alanine, can be measured in CSF when looking for specific conditions. Blood stained CSF is unsuitable. Must be accompanied by paired plasma Li heparin sample (fluoride acceptable). Send by first class post.

Amino Acids in hair protein (*For the diagnosis of Trichothiodystrophy Syndrome only*)
50 mg of hair sample required. Please contact laboratory prior to request.

Ammonia
µmol/L; Take at least 1.0 mL venous or arterial blood into a Li heparin tube standing in ice. Centrifuge at 4°C within 30 minutes. Store plasma at -70°C. Send to this laboratory still frozen (dry ice); neonate up to 100, infant, child, adult up to 50 µmol/L.

B

Bile Salts, profile (*For the diagnosis of bile acid biosynthesis disorders*)
1 mL venous blood in Li heparin tube, separate and send 0.5 ml plasma, or 5 mls urine in a plain tube, contact the laboratory before collection. Send by first class post. Interpretation given with report.
NB. For the diagnosis of bile acid synthesis disorders (this is NOT the same as total bile acid/bile salts quantitation i.e. not for cholestasis in pregnancy).

Biotinidase
u/L; 1 mL venous blood in a Li heparin tube, separate and send 0.5 mL plasma, store at -20°C. Send by first class post Monday-Thursday with normal packaging. Child, adult 2.5-10.5 u/L.

C

Caffeine
mg/L; 0.5 mL venous or capillary blood in a Li heparin tube, separate and send plasma.; patients on high dose caffeine therapy 27-40 mg/L, also reported in neonates receiving theophylline. Assayed weekly on Tuesday.
Carnitine
µmol/L; 1 mL venous blood in a Li heparin tube, separate and send plasma (serum and fluoride possible) store at -20°C. Send by first class post.
total: 23-60 µmol/L
free 15-53 µmol/L
Urine 5 mL plain sample (must be accompanied by blood sample)
Please note that for virtually all clinical purposes an acylcarnitine profile with free carnitine is a more thorough investigation and likely to provide better clinical information.

Catecholamine metabolites – See VMA & HVA.

Catalase staining of fibroblasts
Skin biopsy or cultured fibroblasts
Arrange with Clinical Chemistry before sending the sample.
Contact Clinical Scientists (bleep 095)

Cholestanol
1 mL Li heparin venous blood, separate and send plasma. (Serum acceptable). For diagnosis of Cerebrotendinous Xanthomatosis. Send plasma by first class post. Normal range 3-16 µmol/L.

Collagen Cross-links
1mL urine (min). Fresh sample, protected from light. If delay in posting sample store at -20°C and send 1st class post. For differential diagnosis of PLOD1 defects (EDS Type VI) only.

D

7-Dehydrocholesterol (For diagnosis of Smith Lemli Opitz Syndrome)
1 mL Li heparin venous blood, separate and send plasma by first class post. 10 mL Amniotic fluid for prenatal diagnosis (contact laboratory prior to collection).
Cholesterol and full sterol profile included in the assay.
Abnormal 7-DHC > 5 µmol/L, Normal 7-DHC <2 µmol/L.

DHAP-AT for peroxisomal biogenesis disorders (Fibroblasts)
Skin biopsy – Arrange with Clinical Chemistry before sample collection.
Contact Duty Clinical Scientist (Bleep 095)
**Dimethylglycine**
1 mL Li heparin venous blood, separate and send plasma. 2 mL urine in a plain tube. For the diagnosis of Dimethylglycine dehydrogenase deficiency.

**E**

**Enzyme diagnosis of inherited metabolic diseases - see page 12**

**F**

**Filipin Staining for Niemann Pick Type C (Fibroblasts)**
Skin biopsy – Arrange with Clinical Chemistry before sample collection. Contact Duty Clinical Scientist (Bleep 095)

**Fish Odour Syndrome** - see Trimethylamine (and Dimethylglycine).

**Free Fatty Acids** - see Intermediary metabolites.

**Fumaric Acid** (*For prenatal diagnosis of Fumarate hydratase deficiency*)
Please contact laboratory before sending sample
5 mL Amniotic fluid.

**Fumarate Hydratase** (Fibroblasts, Amniocytes)
Please contact laboratory prior to sending sample
Skin biopsy or Amniotic fluid.

**G**

**Galactitol**
2 mL Plain Urine.
Interpretation given with the result.

**Galactosaemia screen** (Galactose -1-phosphate uridyl transferase in erythrocytes)
0.5 mL venous or capillary whole blood in a lithium heparin tube; send whole blood first class post, normal packaging. Interpretation given with report.
EDTA samples are unsuitable.

**Glutarate (quantitative)**
(For prenatal diagnosis of glutaric aciduria type 2)
10 mL amniotic fluid
Laboratory MUST be contacted
**Glycine (CSF:Plasma ratio)**

0.5 mL venous blood in a Li heparin tube separate and send plasma together with 0.5 mL CSF sample (no additive required) by first class post Monday-Thursday with normal packaging. Interpretation given with report.

**Glyceral Acid (Chirality)**

Determination of L- or D- Glyceral Aciduria

5 mL random urine sample in a plain tube.

**Glycosaminoglycans (mucopolysaccharides, MPS)**

mg/mmol creatinine; one random urine sample or a 20 mL aliquot from a 24 hour collection in a plain bottle, store urine at -20°C, send first class post, normal packaging. Urine must be adequately concentrated (creatinine over 1.0 mmol/L) for a valid result; 0-4wks, 22.1-40.8; 1m-3m, 9.2-38.8; 3-6m, 11.9-34.5; 6m-1y, 4.2-30.5; 1y-2y, 6.8-21.7; 2y-3y, 9.7-19.5; 3y-5y, 6.2-15.4; 5y-7y, 6.2-12.1; 7y-9y, 4.1-10.8; 9y-11y, 4.5-10.8, 11-13y, 2.8-10.4; 13y-15y, 2.0-7.6; >15y, 1.7-4.4.

**H**

**Hair Protein Amino Acids** - see Amino Acids in hair protein.

**Hexanoylglycine**

2 mL random urine (no preservative), store at -20°C send first class post; Used in the diagnosis of medium chain fatty acid oxidation defects. Normal range 0.1-1.1 µmol/mmol creatinine. 5 mL amniotic fluid. Laboratory **MUST** be contacted.

**HMMA** – See VMA

**Homocysteine (total)**

µmol/L; Collect 3 mL blood into an EDTA or Lithium heparin tube separate within 1 hour, store plasma at -20°C. Send first class post, normal packaging. Patient should be fasting (overnight); child, adult 0-18 male, 0-16 female.

If urgent estimation of free homocysteine is required for diagnostic purposes please contact laboratory for sample requirements.
Homovanillic acid (HVA)
µmol/mmol creatinine; 10 mL aliquot of a 24 hour urine collected into 10 mL HCl 6 mol/L CARE store urine at -20°C. Send first class post, normal packaging. ; infant 4-25, 1-5y 2-15. >5y 2-13.

HVA - see Homovanillic acid.

2-Hydroxy Glutaric Acid (Chirality)
For the diagnosis of D-or L- 2- hydroxy glutaric aciduria - 5 ml urine in a plain tube. Please send evidence of increased excretion of 2- hydroxy glutarate.

3- Hydroxybutyrate – see Intermediary metabolites

4-Hydroxybutyric Acid
2 mL urine, 0.5 mL CSF
5 mL Amniotic Fluid For the prenatal diagnosis of succinic semi aldehyde dehydrogenase deficiency
Please contact laboratory prior to sending sample
Interpretation given with report.

Intermediary metabolites (part of fasting hypoglycaemia screen)
Glucose, lactate, free fatty acids, 3-hydroxybutyrate. 2 ml blood into fluoride oxalate, fluoride EDTA or fluoride heparin separate and send plasma by first class post, normal packaging. Haemolysed samples unsuitable for FFAs

Emergency investigation protocol in cases of suspected hypoglycaemia;
Take 2 mL blood into fluoride oxalate and obtain the first urine sample passed for organic acid analysis.

Isovalerylglucose
(For the prenatal diagnosis of Isovaleric Acidaemia and Glutaric aciduria Type 2)
Laboratory MUST be contacted
10 mL Amniotic Fluid.
L

Lactate, fasting
mmol/L; 0.5 mL venous in a fluoride oxalate tube, separate and send plasma first class post; neonate up to 3.0, child 0.9-1.8, adult 0.6-2.4; CSF lactate 0.2 mL in a fluoride oxalate tube, send first class post; neonate up to 3.0, child 0.9-1.8, adult 0.6-2.4 mmol/L.

D,L, lactate (determination of chirality)
Random urine 5 mL. Contact laboratory prior to request.

Long-chain fatty acids - see very long-chain fatty acids.

M

Methotrexate
$10^{-6}$ µmol/L (or similar notation depending on the concentration); 0.75 mL venous or capillary blood in a Li heparin tube; contact laboratory before collection; expected level in a child or adult 48 hours after last dose 1-2 $10^{-6}$ mol/L.

Methylcitric acid
for the prenatal diagnosis of Propionic Acidaemia 10 mL amniotic fluid - laboratory must be contacted prior to collection.

Methylmalonate
5 mL random urine (no preservative). Child normal range 1-8 µmol/mmol creatinine. Adult range 0.2-2.4 µmol/mmol creatinine.
Send urine first class post, normal packaging.

5 mL amniotic fluid (for prenatal diagnosis of methylmalonic acidaemia) laboratory must be contacted before sending sample.

Molecular Genetics see page 13

Mucopolysaccharides (MPS) - see Glycosaminoglycans.
**Muscle Biopsy** - Arrange with Clinical Chemistry before sample collection
Contact Duty Clinical Scientist (Bleep 095)

N
O

**Organic acids, Profile**
10 mL aliquot of a random or a 24 hour urine in a plain bottle, qualitative report given. Boric acid in urine makes analysis impossible—do not use borate. Send urine first class post.

**Orotic acid**
urine µmol/mmol creatinine; 10 mL aliquot of a random or 24 hour urine in a plain bottle, infant/child/adult <3.5.
Send urine first class post.

**5-Oxoproline** (See Pyroglutamic acid)

P

**Phenylalanine, fasting**
µmol/L; 0.5 mL plasma from venous or capillary blood in a Li heparin tube; newborn 40-110, < 6mo 32-128, 6mo-2y 40-140, 2y-10y 20-130, 10y-17y 30-115, adult 40-100.
dried blood spot (Guthrie card) for screening and monitoring dietary control in PKU, adequate dietary control 200-400 (depending on age).

**Phosphoethanolamine**
µmol/mmol Cr; 10 mL urine in a plain container; child, adult <10 µmol/mmol Cr, for hypophosphatasia - heterozygote 3-8 x normal, homozygote 10-50 x normal.

**Phytanic acid**
1 mL Li heparin blood; separate and send plasma by first class post normal 0.2 -19.3 µmol/L; interpretation given with the report.

**Phytosterols**
1 mL Li heparin venous blood, separate and send plasma (serum acceptable). Send by first class post. Quantitation of campesterol, stigmasterol and sitosterol included.
**Pipecolic Acid**
1 mL Li heparin blood/EDTA/Serum, 0.5 mL CSF, 5 mL urine in plain container. Separate blood and send plasma or serum, first class post. Interpretation given with report.

**Plasmalogens in RBC**
*For the diagnosis of peroxisomal biogenesis disorder and rhizomelic chondrodysplasia punctata.*
2 mL EDTA; red blood cells (RBC), wash RBC x 3 with saline, send by first class post. Please phone the laboratory before sending.; interpretation given with the report.

**Pristanic Acid**
1 mL Li heparin, separate and send plasma by first class post. Included in Very Long Chain Fatty Acid Assay. Normal 0-1.88 µmol/L.

**Pyruvate, with lactate, fasting**
Special arrangements are required for this analyte. Please contact Duty Clinical Scientist prior to collection (Bleep No 095).

**Pyridinoline/deoxypyridinoline**
5 mls fresh urine for assessment of collagen/bone turnover.

**Pyroglutamic Acid**
*For the prenatal diagnosis of Glutathione Synthetase deficiency*  
Laboratory **MUST** be contacted before sending sample.
5 mL Amniotic Fluid.

**Reducing Substances**
- Urine: 5 mL aliquot from a fresh random specimen  
  - glucose, pH and reducing substance (by benedicts)
- Faeces: Analysis only carried out following discussions with Dr Olpin/Duty Biochemist
Sitosterol – see Phytosterols

Skin Biopsy – A protocol is available from the laboratory describing a suggested technique and transport arrangements (Tel 0114 2717267) Tissue culture medium and consent forms are available from the laboratory and samples should be arranged in advance to arrive not later than 4.30pm. A completed consent form should accompany all samples

Sterols, Profile – See 7-dehydrocholesterol (Included in 7-DHC Test)

Sweat test – please contact the laboratory

Thiosulphate
Urine mmol/mmol creatinine; 10 mL random urine sample send by first class post. Please include drug history at time of sample (particularly antibiotics). Interpretation given with the report.

Trimethylamine (And Oxide)
For the diagnosis of primary and secondary trimethylaminuria (Fish Odour syndrome)
24 hour urine collected in HCl is optimal (10 mL of 6N HCl)
20 mL random urine is acceptable if fresh sample but please acidify to pH1 with HCl before sending by first class post

TMA 2.5-10.9 µmol/mmol crt  TMA Oxide 17.0-147.0 µmol/mmol crt
TMA: TMA oxide ratio 0.05-0.21
Very long-chain fatty acids (peroxisomal disorders)
1 mL venous blood in a Li heparin tube or EDTA send plasma by first class post. (serum also accepted); µmol/L C₂₂ 30-112, C₂₄ 14-80, C₂₆ 0.33-1.50, C₂₄/C₂₂ 0.44-0.97, C₂₆/C₂₂ 0.006-0.028; Assay includes pristanic and phytanic acid quantitation. Fibroblast; Amniocyte; chorionic villus cell assays. Please contact laboratory.

VMA (HMMA, vanillylmandelic acid, VMA)
µmol/mmol creatinine; 10 mL aliquot of a 24 hour urine collected into plastic bottle with 10 mL HCl 6 mol/L CARE, store urine at -20°C. Send first class post, normal packaging.; infant 2-12, 1-5y 2-9 > 5y 1-7
<table>
<thead>
<tr>
<th>Test</th>
<th>Cost (£)</th>
<th>Cost to East Midlands and South Yorkshire</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acylcarnitine Profile</td>
<td>66.00</td>
<td>52.50</td>
</tr>
<tr>
<td>Amino acid in hair protein</td>
<td>184.66</td>
<td>92.33</td>
</tr>
<tr>
<td>Amino Acids: Full Quantitation</td>
<td>78.25</td>
<td>78.25</td>
</tr>
<tr>
<td>Part Quantitation</td>
<td>56.33</td>
<td>56.33</td>
</tr>
<tr>
<td>TLC of Urine (includes spot test and DMB test for MPS)</td>
<td>61.00</td>
<td>61.00</td>
</tr>
<tr>
<td>Bile salts in plasma or urine (Quantitation of Taurocholic acid)</td>
<td>48.72</td>
<td>24.36</td>
</tr>
<tr>
<td>Biotinidase</td>
<td>51.60</td>
<td>51.60</td>
</tr>
<tr>
<td>Carnitine (Total and Free)</td>
<td>86.09</td>
<td>86.09</td>
</tr>
<tr>
<td>Cholestanol in plasma</td>
<td>90.00</td>
<td>45.00</td>
</tr>
<tr>
<td>Collagen Cross-links</td>
<td>80.00</td>
<td>80.00</td>
</tr>
<tr>
<td>7-dehydrocholesterol in plasma</td>
<td>90.00</td>
<td>45.00</td>
</tr>
<tr>
<td>Dimethylglycine</td>
<td>48.72</td>
<td>24.36</td>
</tr>
<tr>
<td>Galactitol</td>
<td>88.63</td>
<td>44.32</td>
</tr>
<tr>
<td>Galactose 1 Phosphate uridyl transferase</td>
<td>34.42</td>
<td>34.42</td>
</tr>
<tr>
<td>Glutaric acid (in cerebrospinal fluid)</td>
<td>157.24</td>
<td>78.62</td>
</tr>
<tr>
<td>Hexanoylglycine in urine</td>
<td>157.24</td>
<td>78.62</td>
</tr>
<tr>
<td>HVA/VMA</td>
<td>75.00</td>
<td>75.00</td>
</tr>
<tr>
<td>4-hydroxybutyrate</td>
<td>157.24</td>
<td>78.62</td>
</tr>
<tr>
<td>Intermediary metabolites</td>
<td>51.60</td>
<td>51.60</td>
</tr>
<tr>
<td>Free Fatty Acids</td>
<td>17.20</td>
<td>17.20</td>
</tr>
<tr>
<td>β hydroxybutyrate</td>
<td>17.20</td>
<td>17.20</td>
</tr>
<tr>
<td>Methylmalonic acid in urine</td>
<td>70.44</td>
<td>35.22</td>
</tr>
<tr>
<td>Mucopolysaccharide electrophoresis</td>
<td>103.19</td>
<td>103.19</td>
</tr>
<tr>
<td>Organic Acid (GCMS) including extraction, assay and identification</td>
<td>88.30</td>
<td>44.15</td>
</tr>
<tr>
<td>Organic Acids (GCMS) (Peak identification only sent with a copy of the chromatogram)</td>
<td>40.70</td>
<td>20.35</td>
</tr>
<tr>
<td>Orotic Acid</td>
<td>75.00</td>
<td>37.50</td>
</tr>
<tr>
<td>pH and reducing substances</td>
<td>17.20</td>
<td>17.20</td>
</tr>
<tr>
<td>Phytosterols</td>
<td>90.00</td>
<td>45.00</td>
</tr>
<tr>
<td>Pipecolic Acid (in plasma/serum/urine/CSF)</td>
<td>157.24</td>
<td>78.62</td>
</tr>
<tr>
<td>Plasmalogens</td>
<td>157.24</td>
<td>78.62</td>
</tr>
<tr>
<td>Pyruvate</td>
<td>40.00</td>
<td>40.00</td>
</tr>
<tr>
<td>Sulphocysteine</td>
<td>70.44</td>
<td>70.44</td>
</tr>
<tr>
<td>Thiosulphate</td>
<td>40.00</td>
<td>40.00</td>
</tr>
<tr>
<td>Total Homocysteine</td>
<td>34.42</td>
<td>34.42</td>
</tr>
<tr>
<td>Trimethylamine (and oxide) in urine to include Dimethylglycine follow-up if TMA is normal</td>
<td>134.00</td>
<td>67.00</td>
</tr>
<tr>
<td>Very long chain fatty acids and phytanic &amp; pristanic acids (combined) in plasma</td>
<td>82.00</td>
<td>41.00</td>
</tr>
<tr>
<td>Additional charge for analyses out of hours</td>
<td>80.00</td>
<td>40.00</td>
</tr>
<tr>
<td>Test</td>
<td>Cost (£)</td>
<td>Cost to East Midlands and South Yorkshire</td>
</tr>
<tr>
<td>-------------------------------------------</td>
<td>----------</td>
<td>------------------------------------------</td>
</tr>
<tr>
<td>Quantitation in amniotic fluid for prenatal diagnosis</td>
<td></td>
<td></td>
</tr>
<tr>
<td>4 hydroxybutyrate</td>
<td>475.00</td>
<td>237.50</td>
</tr>
<tr>
<td>7 Dehydrocholesterol</td>
<td>350.00</td>
<td>175.00</td>
</tr>
<tr>
<td>Fumaric acid</td>
<td>350.00</td>
<td>175.00</td>
</tr>
<tr>
<td>Glutaric acid</td>
<td>350.00</td>
<td>175.00</td>
</tr>
<tr>
<td>Isovalerylglycine</td>
<td>410.00</td>
<td>205.00</td>
</tr>
<tr>
<td>Methylcitrate</td>
<td>350.00</td>
<td>175.00</td>
</tr>
<tr>
<td>Methylmalonic acid</td>
<td>350.00</td>
<td>175.00</td>
</tr>
<tr>
<td>Pyroglutamic acid (5-oxoproline)</td>
<td>475.00</td>
<td>237.50</td>
</tr>
<tr>
<td>Determination of chirality (in urine)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Glyceric acid, or 2-hydroxyglutaric acid</td>
<td>375.00</td>
<td>187.50</td>
</tr>
</tbody>
</table>

**NOTES**

a) Please give advance warning of prenatal diagnoses. These are priced on the assumption that the result is required urgently. Control amniotic fluid samples are run in parallel and should ideally be submitted by the requesting centre with the sample from the at-risk pregnancy.

<table>
<thead>
<tr>
<th>Test</th>
<th>Cost (£)</th>
<th>Cost to East Midlands and South Yorkshire</th>
</tr>
</thead>
<tbody>
<tr>
<td>Routine Chemistry</td>
<td>11.50</td>
<td>11.50</td>
</tr>
<tr>
<td>Ammonia</td>
<td>17.20</td>
<td>17.20</td>
</tr>
<tr>
<td>Lactate</td>
<td>17.20</td>
<td>17.20</td>
</tr>
<tr>
<td>Caffeine</td>
<td>27.00</td>
<td>27.00</td>
</tr>
<tr>
<td>Cyclosporin</td>
<td>30.00</td>
<td>30.00</td>
</tr>
<tr>
<td>Methotrexate</td>
<td>30.00</td>
<td>30.00</td>
</tr>
<tr>
<td>Tacrolimus</td>
<td>30.00</td>
<td>30.00</td>
</tr>
<tr>
<td>Sweat test (collection and analysis)</td>
<td>55.00</td>
<td>55.00</td>
</tr>
<tr>
<td>Additional charge for analyses out of hours</td>
<td>80.00</td>
<td>40.00</td>
</tr>
</tbody>
</table>
The following tests are free of charge for patients living in districts covered by the East Midlands and South Yorkshire Newborn Screening Contracts

<table>
<thead>
<tr>
<th>Test</th>
<th>Cost (£)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Tissue culture</strong></td>
<td></td>
</tr>
<tr>
<td>Establishing a primary culture from skin biopsy (or recovered cryopreserved skin sample), growing on under sterile conditions (mycoplasma-free), cryopreservation and retention for at least 2 years</td>
<td>250.00</td>
</tr>
<tr>
<td>Growing on of fibroblast cell line for assay when received as cells from outside laboratory</td>
<td>140.00</td>
</tr>
<tr>
<td>Cryopreservation of skin samples for later culture if required (retention period not less than 6 months)</td>
<td>50.00</td>
</tr>
<tr>
<td>Recovery of cryopreserved cultured cells and growing on</td>
<td>65.00</td>
</tr>
<tr>
<td>Onward dispatch of living culture (carriage extra at cost); within the UK</td>
<td>50.00</td>
</tr>
<tr>
<td>Abroad</td>
<td>90.00</td>
</tr>
<tr>
<td><strong>Assays on cultured cells</strong></td>
<td></td>
</tr>
<tr>
<td>$^{14}$CO$_2$ or $^{14}$C-incorporation assays with cultured fibroblasts (propionic, isovaleric etc)</td>
<td>310.00</td>
</tr>
<tr>
<td>3-Methylcrotonyl-CoA carboxylase (fibroblasts)</td>
<td>311.60</td>
</tr>
<tr>
<td>Carnitine palmitoyl transferase Type I – Cultured Fibroblasts</td>
<td>491.66</td>
</tr>
<tr>
<td>Carnitine palmitoyl transferase Type II – Cultured Fibroblasts</td>
<td>350.00</td>
</tr>
<tr>
<td>Carnitine acylcarnitine Translocase (CATR)</td>
<td>450.00</td>
</tr>
<tr>
<td>Carnitine Transporter assay (fibroblasts)</td>
<td>240.00</td>
</tr>
<tr>
<td>Catalase staining</td>
<td>256.00</td>
</tr>
<tr>
<td>Citrulline incorporation into fibroblasts for detection of defects of argininosuccinate synthase and argininosuccinate lyase</td>
<td>370.00</td>
</tr>
<tr>
<td>Cultured in low biotin medium for above</td>
<td>590.00</td>
</tr>
<tr>
<td>DHAP-AT</td>
<td>460.80</td>
</tr>
<tr>
<td>Fatty acid oxidation screen (tritiated myristate palmitate and oleate in parallel on cultured fibroblasts)</td>
<td>275.00</td>
</tr>
<tr>
<td>Fibroblast acylcarnitine profile for the characterisation of fatty acid oxidation defects, As a lone request</td>
<td>275.00</td>
</tr>
<tr>
<td>Fibroblast tritiated fatty acid oxidation flux plus fibroblast acylcarnitine profiles</td>
<td>350.00</td>
</tr>
<tr>
<td>Filipin stain</td>
<td>256.00</td>
</tr>
<tr>
<td>Fumarate hydratase</td>
<td>460.25</td>
</tr>
<tr>
<td>Glutamate dehydrogenase (fibroblasts)</td>
<td>320.00</td>
</tr>
<tr>
<td>Incorporation of phenylalanine L[ring 2, 6-3H] and L-Ornithine [5-14C into – Cultured Fibroblasts for the detection of patients with hyperornithinaemias (HHH &amp;OAT) Cultured Fibroblasts</td>
<td>365.24</td>
</tr>
<tr>
<td>Long- and short-chain 3-hydroxyacyl-CoA dehydrogenases</td>
<td>400.00</td>
</tr>
<tr>
<td>Ornithine Oxo-Acid Aminotransferase assay (EC 2.6.1.13) for the confirmation of OAT – Cultured Fibroblasts</td>
<td>370.00</td>
</tr>
<tr>
<td>Propionyl CoA carboxylase (fibroblasts)</td>
<td>320.00</td>
</tr>
<tr>
<td>Test</td>
<td>Cost (£)</td>
</tr>
<tr>
<td>-------------------------------------------------------------</td>
<td>----------</td>
</tr>
<tr>
<td>Pyruvate Carboxylase</td>
<td>311.60</td>
</tr>
<tr>
<td>Very long chain fatty acids in cultured cells (fibroblasts, amniotic fluid cells or chorionic villus cells)</td>
<td>335.00</td>
</tr>
<tr>
<td>Very long-chain acyl-CoA dehydrogenase (ferricenium linked assay)</td>
<td>400.00</td>
</tr>
</tbody>
</table>

**NOTES**

a) Cultured fibroblasts submitted for assay **MUST** be mycoplasma-free. If your local cytogenetics laboratory cannot ensure this then please send skin biopsy directly to us for culture. Cultures that are infected when received will be discarded.

b) The prices shown are indicative only. For some assays there are substantial reductions for multiple samples received at one time e.g. for family studies.

c) For prenatal diagnoses using cultured amniotic fluid cells or chorionic villus, reference material, cultured under the same conditions as the suspect sample, should be supplied by the referring centre. The above prices are increased by 40% for prenatal diagnosis or 70% if recovered cryopreserved material (e.g. the index case) is included as a positive control.

d) The prices shown do not include VAT, not usually applicable within the NHS. It may be applied to users outside the NHS dependent on current VAT rules.