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| --- | --- | --- | --- | --- |
| **Patient information**  Please tick if patient is **not** eligible for NHS treatment 🖵 | |  | **Referring clinician / Laboratory information**  Please complete in full or the report may be delayed | |
| Surname |  |  | Consultant |  |
| Forename |  |  | Department |  |
| Date of birth |  |  | Contact number |  |
| Sex |  |  | Email \* |  |
| NHS Number |  |  | **N.B.** reports can be sent to nhs.net email addresses or encrypted to other addresses with prior arrangement | |
| Hospital no. |  |  | Address: |  |
| Your lab no. |  |  |  |  |
| Address |  |  |  |  |
| Postcode |  |  | Postcode |  |

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| **Sample requirements (see page 2 for available tests)**  **N.B.** DO NOT spin / separate / freeze blood samples! | |
| Galactosaemia (galactose-1-phopshate uridyltransferase) | 0.5 ml blood in lithium heparin (EDTA also OK). 1st class post |
| Galactokinase | 2 ml blood in lithium heparin. Sample must arrive within 24 hours of collection and lab must be contacted before arranging testing |
| Lysosomal enzymes (‘white cell enzymes’) | 5 to 10 ml blood in lithium heparin  To arrive at our laboratory within 24 hours of collection.  Courier is preferable to guaranteed next day delivery. |
| Blood for single enzyme testing  e.g. Fabry disease, Pompe disease |
| Urine glycosaminoglycans (GAGs) / mucopolysaccharides | 10 to 20 ml urine without preservative. 1st class post  Store frozen if not sent immediately. |
| Urine oligosaccharides |
| Skin biopsy (for culture, storage and / or testing) | Collect into sterile culture medium containing fetal bovine serum  (or sterile saline if culture medium not available) |
| Cultured cells or prenatal samples  (fibroblasts, amniocytes, chorionic villi) | Please contact the laboratory |
| Liver biopsy (for OTC / CPS1 testing) |
| Tay-Sachs carrier testing (biochemical) | 8 to 10 ml lithium heparin blood from all patients  5 ml clotted blood if patient is not pregnant  To arrive at our laboratory within 24 hours of collection. |
| **Please do not use this form for DNA test requests – a separate genetics request form is available** | |

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| **Test Requested**  Please provide as much clinical, biochemical or family information as possible |
| Is this test urgent? 🖵 If so, please state why |

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| **Sample information** | |
| Date taken: | Time taken: |

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| Urea Cycle Disorders | Test |
| Citrullinaemia\* | Citrulline incorporation |
| Argininosuccinic aciduria\* |  |
| Carbamyl phosphate synthetase deficiency\* (CPSI) | Carbamyl phosphate synthetase |
| Ornithine transcarbamylase deficiency (OTC)\* | Ornithine transcarbamylase |
|  |  |
| Mitochondrial carboxylases | Test |
| Propionic acidaemia\* | Propionyl-CoA carboxylase |
| Pyruvate carboxylase deficiency\* | Pyruvate carboxylase |
| 3-methylcrotonyl-CoA carboxylase deficiency\* | 3-methylcrotonyl-CoA carboxylase |
|  |  |
| Mucopolysaccharidosis | Test |
| MPS type 1 Hurler / Scheie | α-iduronidase |
| MPS type 2 Hunter | iduronate sulphatase |
| MPS type 3A Sanfilippo A | Heparan sulphamidase |
| MPS type 3B Sanfilippo B | α-N-acetylglucosaminidase |
| MPS type 3C Sanfilippo C | Glucosamine N-acetyl transferase |
| MPS type 3D Sanfilippo D | Glucosamine N-acetyl-6-sulfatase |
| MPS type 4A Morquio A | N-acetylgalactosamine 6-sulphatase |
| MPS type 4B Morquio B | β-galactosidase |
| MPS type 6 Maroteaux Lamy | Arylsulphatase B |
| MPS type 7 Sly | β-glucuronidase |
| Multiple sulphatase deficiency | Various sulphatase enzymes |
|  |  |
| Neuronal ceroid lipofuscinosis (NCL) / Batten disease | Test |
| Infantile neuronal ceroid lipofuscinosis (INCL),  NCL type 1 | Palmitoyl protein thioesterase |
| Late-infantile neuronal ceroid lipofuscinosis (LINCL),  NCL type 2 | Tripeptidyl peptidase I |
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| Galactose metabolism | Test |
| Classic galactosaemia | Galactose 1-phosphate uridyltransferase |
| Galactokinase deficiency | Galactokinase |
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| Tay-Sachs carrier testing | |
| Hexosaminidase by heat inactivation | |
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| Glycosaminoglycans, Oligosaccharides | Test |
| Glycosaminoglycans (GAG) / Mucopolysaccharides | Quantitative & electrophoresis |
| Oligosaccharides | oligos by TLC |
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| Lysosomal enzyme panels | |
| Neurodegenerative (blood) | 15 tests incl. chitotriosidase,  Krabbe disease, Metachromatic leukodystrophy, PPT, TPP1 |
| Hepato(spleno)megaly (blood) | 14 tests incl. chitotriosidase, Niemann Pick A/B, LAL deficiency, Gaucher disease |
| Fetal hydrops (amniotic fluid) | Up to 12 tests depending on sample / requirements incl. GAGs, MPS7, I-cell, Niemann Pick A/B & C (filipin staining), Gaucher disease |
|  | |
| Other disorders | Test |
| N-acetyl neuraminidase | Sialidosis |
| I-cell disease &  mucolipidosis III | Multiple hydrolases |
| α-fucosidase | Fucosidosis |
| α-mannosidase | α-mannosidosis |
| β-mannosidase | β-mannosidosis |
| Schindler disease | α-N-acetylgalactosaminidase |
| Aspartylglycosaminuria | Aspartylglucosaminidase |
| Tay-Sachs and  Sandhoff diseases | Hexosaminidase A and B |
| GM1-gangliosidosis | β-galactosidase |
| Metachromatic leucodystrophy | Arylsulphatase A |
| Krabbe disease | Galactocerebrosidase |
| Gaucher disease | β-glucosidase (glucocerebrosidase) & chitotriosidase |
| Chitotriosidase only | Chitotriosidase |
| Niemann-Pick disease  types A & B | Sphingomyelinase |
| Wolman disease / Cholesterol ester storage disease  Lysosomal acid lipase (LAL) deficiency | acid esterase  lysosomal acid lipase |
| Niemann-Pick disease type C | Filipin staining of stored cholesterol |
| Pompe disease  Glycogen storage disease (GSD) type 2 | α-glucosidase  acid maltase |
| Zellweger syndrome and other generalised peroxisomal disorders\* | Dihydroxyacetone phosphate acyltransferase (DHAPAT) |
| Maple syrup urine disease\* (MSUD) | branched chain ketoacid decarboxylase |
| Fabry disease | α-galactosidase |
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| \* Test not currently covered by our UKAS accreditation / scope.  Please contact the laboratory for further information. | |

Please see our website for sample requirements for specific tests:

[www.viapath.co.uk/departments-and-laboratories/biochemical-genetics-laboratory-at-guys](http://www.viapath.co.uk/departments-and-laboratories/biochemical-genetics-laboratory-at-guys)