**Exeter Exome Sequencing Request Form**

Sample requirements: Please send at least ***10µg*** *DNA or**EDTA blood to: Prof. S. Ellard, Molecular Genetics Laboratory, RILD level 3, Royal Devon & Exeter NHS Foundation Trust, Barrack Road, Exeter EX2 5DW* [rde-tr.MolecularGeneticsAdmin@nhs.net](mailto:rde-tr.MolecularGeneticsAdmin@nhs.net)

* We welcome enquiries – please contact Dr Júlia Baptista ([Julia.baptista@nhs.net](mailto:Julia.baptista@nhs.net) or 01392 408247) and Dr Emma Baple (ebaple@nhs.net or 01392 405749) to discuss cases
* Prices include co-segregation analysis and confirmatory testing by Sanger sequencing or droplet digital PCR (for CNVs)
* We use the Agilent capture technology which can detect partial/whole gene deletions in addition to base substitutions and small insertions/deletions
* Non-NHS referrals will incur a 25% surcharge

|  |  |  |  |
| --- | --- | --- | --- |
| **CLINICIAN DETAILS** | | | |
| CLINICIAN NAME: | | TELEPHONE: | |
| E-MAIL ADDRESS FOR REPORT (nhs.net): | | LABORATORY EMAIL ADDRESS FOR REPORT (nhs.net): | |
| **PEDIGREE & CLINICAL DETAILS** | | | |
| **PLEASE PROVIDE A PEDIGREE AND FULL DETAILS OF THE CLINICAL PHENOTYPE, DIAGNOSES UNDER CONSIDERATION AND PRIOR INVESTIGATIONS (GENETIC AND NON-GENETIC, FOR EXAMPLE, NEUROIMAGING) TO AID THE INTERPRETATION OF VARIANTS. WHERE POSSIBLE PLEASE SEND THE MOST RECENT SUMMARY CLINIC LETTER WITH THE EXOME REQUEST FORM**: | | | |
| ETHNIC ORIGIN: | CLINICAL GENETICS NUMBER: | | PLEASE TICK IF URGENT |

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **WHICH EXOME TEST? (for further information see** [http://www.exeterlaboratory.com/test/exome-sequencing-services](http://www.exeterlaboratory.com/test/exome-sequencing-services/)**)** | | | | | | | |
| **Trio analysis**   £1,995  *23, 244 genes (Agilent v6 exome)*  Exome sequencing for an affected proband and their unaffected parents to identify *de novo* heterozygous, compound heterozygous, homozygous or X-L recessive disease-causing variants. | | | **Affected sib pair analysis in consanguineous families**  £1,595 *23,244 genes (Agilent v6 exome)*    Exome sequencing for two affected siblings followed by co- segregation analysis of likely disease-causing variants in the parents and additional affected or unaffected siblings. A homozygosity analysis is performed to identify autozygous regions. | | | | |
| **Couple analysis for lethal fetal disorders**  £1,595  *23, 244 genes (Agilent v6 exome)*  Exome sequencing for a couple who have had one or more pregnancies affected with a lethal, presumed autosomal recessive disorder (Ellard *et al* 2015 Eur J Hum Genet [PMID 24961629](http://www.ncbi.nlm.nih.gov/pubmed/?term=24961629)). Our strategy identifies genes where both parents have a heterozygous potentially pathogenic variant. Likely disease-causing variants are then tested in the affected fetus(es) and any unaffected siblings to confirm co-segregation. This approach conserves precious fetal samples and/or is appropriate for cases where fetal DNA is of insufficient quality or quantity for exome sequencing.  **Price:**  £1650: “whole” exome (23,243genes)\*  £1320: rare disease exome  (6,110 genes) \*  This strategy is most likely to yield a diagnosis for unrelated couples with multiple affected fetuses but has been successful for couples with a single affected pregnancy or who carry the same heterozygous disease-causing variant. | | | | | | | |
| **Bespoke virtual gene panel**  £850-£1,300  *Price according to gene panel size and exome capture (6,110 known disease genes or 23, 244 genes “whole” exome)*  This is the only option for some families due to sample availability or pedigree structure. A virtual gene panel can either be provided by the clinician, a bespoke design based on phenotype (we use a combination of genes selected by HPO and OMIM search terms), a panel described elsewhere (research publication or non-NHS laboratory service) or a combination of these. The virtual gene panel may include a single gene or many hundreds.  PLEASE E-MAIL THE EXETER EXOME TEAM ([rde-tr.MolecularGeneticsAdmin@nhs.net](mailto:rde-tr.MolecularGeneticsAdmin@nhs.net)) TO DISCUSS BESPOKE VIRTUAL GENE PANEL TESTS | | | | | | | |
| **DNA SAMPLES (PLEASE ENSURE NAMES ARE INCLUDED ON THE PEDIGREE)** | | | | | | | |
| SURNAME: | FORENAME: | D.O.B: | | NHS NUMBER: | GENDER: | DECEASED | AFFECTED UNAFFECTED |
| SURNAME: | FORENAME: | D.O.B: | | NHS NUMBER: | GENDER: | DECEASED | AFFECTED UNAFFECTED |
| SURNAME: | FORENAME: | D.O.B: | | NHS NUMBER: | GENDER: | DECEASED | AFFECTED UNAFFECTED |