**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

* We welcome enquiries – please contact Karen Stals (karen.stals@nhs.net or 01392 408249) 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

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| **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 [ ]  |

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| **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) 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 [ ]  |
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