University of Iowa
Stead Family
Children's Hospital

Shivanand R. Patil Cytogenetics & Molecular Laboratory

200 Hawkins Drive, W101GH
Iowa City, IA 52242
319-356-3877
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Exome Sequencing Informed Consent

- **What is the purpose of Exome Sequencing?**
  The exome is the most important region within the human genome and is the DNA "code" responsible for making most proteins. Many genetic changes that cause medical conditions are found in the exome. The purpose of Exome Sequencing it to identify changes in the DNA code, also referred to as variants, that may be contributing to you or your child's condition.
  DNA will be extracted from the specimen that you provide. A machine will generate data that represents the sequence of your DNA. Information from your clinic visit will be combined with this data to determine if any genetic changes may be the cause of a medical condition.

- **What types of results are reported?**
  - Positive: A genetic variant has been identified that is known to cause the condition that was described in the clinical referral
  - Negative: No causative genetic variants were identified based on the available scientific evidence at the time of referral.
  - Variant of Uncertain Significance: A variant has been identified that lacks definitive evidence that it is causal of the indicated clinical condition. The change occurs in a gene known to be associated with the individual's clinical findings, but the specific change has either not been identified in other individuals, or it is unclear how the change affects the gene. Therefore, we are unable to determine if the variant is the explanation for the clinical features seen in the patient.

- **What are secondary findings?**
  Secondary findings are not related to your medical condition but can indicate a previously undiagnosed serious conditions that has the potential to be prevented or treated if diagnosed.
  The American College of Medical Genetics and Genomics (ACMG) is a professional organization that maintains guidelines about when it is medically appropriate to offer secondary findings. University of Iowa Hospitals and Clinics follows these guidelines. You have the option to received known pathogenic or likely pathogenic variants in the genes recommend by ACMG as part of this test. For a full list of genes recommended by the ACMG Secondary Findings v3.1 see Miller et al. Genet Med. 2022 (PMID: 35802134).

- **What types of results are not included in the report?**
  Sequence variation is normal and present in everyone's genome. Sequence variants classified during analysis as 'benign' or 'likely benign', will not be reported.
  Autosomal recessive conditions require *two* copies of the same gene to have a pathogenic variant to result in the condition. For autosomal recessive conditions, the presence of only *one* of these variants is typically not the cause of a genetic condition. This is known as being a "carrier" for a genetic condition. Carrier status for autosomal recessive conditions, if identified in genes unrelated to the reason for referral, is not reported.

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- **What are the risks associated with Exome Sequencing?**
  Exome Sequencing may identify serious and untreatable conditions about you or your family. This may cause increased psychological stress. Exome Sequencing may identify unexpected information about family relationships (e.g. misattributed parentage or consanguinity). The laboratory will contact your provider if it is believed that misattributed parentage has been identified in trio analysis. Genetic counseling is recommended.
  The Genetic Information Nondiscrimination Act is a federal law that protects individuals from discriminatory employment hiring practices and health insurance coverage. The law, however, has limitations (e.g. disability, long-term care, or life insurance coverage).

- **What are the limitations associated with Exome Sequencing?**
  Exome Sequencing is highly accurate way of looking at thousands of protein-coding genes with one test. However, this test cannot detect all possible genetic changes. Interpretation of exome data is based on current scientific knowledge which may change over time. Scientific knowledge is constantly growing, and your care provider may request re-analysis in certain circumstances. As new knowledge is discovered, interpretation of this result may change. The laboratory does not report out changes in interpretation of variants automatically but may issue an updated report if requested by the provider. A negative result does not indicate the absence of a genetic cause
  This test is not designed to detect the following:
    - Variation in non-coding sequences
    - Chromosome rearrangements
    - Copy number variants
    - Trinucleotide repeat expansions
    - Mitochondrial variants

  Additionally, human error can occur. Events such as specimen contamination or sample mix-up are highly unlikely but could yield an incorrect result. Numerous laboratory regulations and procedures exist to mitigate this risk to patient safety.

- **How and when will I receive my Exome Sequencing results?**
  Generally, results will be communicated to your provider in about four weeks. These results will be viewable in MyChart. Your provider will discuss these results with you in further detail.
  The University of Iowa offers genetic counseling before and after Exome Sequencing. It is highly recommended to pursue genetic counseling before and after Exome Sequencing results are issued.

- **Who has access to my Exome Sequencing data?**
  Only laboratory personnel will have access to individually identifiable patient data or specimens. After testing is complete, **deidentified** data or remanent specimen may be used for quality control or research purposes. De-identified data may also be submitted to healthcare databases.

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Patient Information
Name DOB:

- **Patient consent**
  I have read or have had read to me the Exome Sequencing Informed Consent document and understand the information regarding this testing.

- **Patient or guardian's signature/date:**

_____Date_____

Yes, include secondary findings, if any, as part of my Exome Sequencing report:

_____(Patient or Guardian's Initials)

No, do not include secondary findings as part of my Exome Sequencing report:

_____(Patient of Guardian's Initials)

- **Healthcare provider's signature/date:**

_____Date_____

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Family member Information
Name: DOB: Relationship to proband:

- **Participating family member's consent:**
  Family member samples are used to help evaluate the results obtained on the person being tested. Only inheritance information for variants identified in the patient will be reported. Family members may opt out of having inheritance information reported for ACMG secondary findings genes if identified in the patient. In the event that a secondary finding is also identified in a family member, a separate report will be issued to that family member if requested below.

  I have read or have had read to me the Exome Sequencing Informed Consent document and understand the information regarding this testing.

  Family Member/guardian Signature:

  _____Date_____

  Include my status in the patient report and issue a separate report to me if I share ACMG secondary findings gene variants identified in the patient:

  _____(Initials)

  Do not include my status in the patient report nor issue a separate report if I share ACMG secondary findings gene variants identified in the patient:

  _____(Initials)

- **Healthcare provider's signature/date:**

  _____Date_____

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