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ECGI Copy

ExomSure | Dx MendeliomSure | Dx PanelSure | NGS

I, (name) _____, voluntarily request Eurofins Clinical Genetics India Pvt.Ltd. (ECGI) to perform **ExomSure I Dx** / **MendeliomSure I DX** / **PanelSure I Dx** sequencing in myself/my child (child's name) _____.

Given the complexity of the exome analysis, **genetic counseling and informed consent by a trained medical geneticist or genetic counselor is required prior to and after undergoing this testing.** Informed consent is a process that provides education about genetics, and the options, benefits, limitations, and consequences of genetic testing. Genetic counseling provides the patient with informed consent prior to the decision to undergo testing and with the opportunity to review the results of the test in detail.

What is the ExomSure | Dx ?

- The ExomSure I Dx is ECGI's whole exome sequencing (WES) test. This test only targets the region of the genome that contains the exons of the genes, called the exome. The exome is estimated to comprise approximately 1-2% of the genome, yet contains approximately 85% of disease-causing pathogenic variants.
- This test may detect variants in known disease-associated genes or variants in genes that have not yet been associated with disease. Only variants in genes associated with disease will be reported. Based on studies, WES is expected to provide a diagnosis in 20-30% of the cases for rare and ultra-rare disorders.

What is the MendeliomSure | Dx ?

- MendeliomSure I Dx only covers genes involved in Mendeliome disease.

Why are parental samples needed?

- In order to interpret your results, other family members may also need to have WES or targeted testing depending on the family history of the disorder and whichmembers are available for testing. ECGI, in consultation with your ordering physician, can recommend which other family members need to be tested.

How is the ExomSure | Dx MendeliomSure | Dx PanelSure | NGS performed?

- This test requires 5-10 ml of blood from which DNA will be extracted. Blood draw can have risks associated with it, such as bruising and bleeding. WES is performed using next generation sequencing (NGS) technology. Using an internally developed filtering algorithm, a list of variants identified from the sequencing is generated and analyzed to identify the variants that might explain the patient's phenotype.
- ExomeSure I Dx capture 54 Mb of Exome.
- MedeliomSure I DX capture 22 Mb of Exome.
- PanelSure I Dx capture disease specific genes and sizes capturing varies from panel to panel.

Limitations of WES by NGS

- A fraction of the exome, estimated to be about 10-15% of the exome, will not have sufficient coverage to accurately determine if a pathogenic variant is present. Therefore, pathogenic variants in these regions will not be detected by this analysis.
- NGS cannot accurately sequence repetitive regions, such as trinucleotide repeats. This means that NGS cannot provide data on regions such as the fragile X syndrome repeat region, the Huntington disease repeat region, or the myotonic dystrophy repeat region.
- Results from WES may indicate that additional testing, such as full gene sequencing to fill-in exons with poor coverage or deletion/duplication analysis, is recommended.
- Copy number variation (CNV) is not evaluated in the **ExomSure I Dx / MendeliomSure I Dx / PanelSure I Dx** test.

Potential risks associated with ExomSure I Dx / MendeliomSure I Dx / PanelSure I Dx :

- Pathogenic variants in genes that lead to conditions for which the patient currently has no features may be discovered (such as cancer, neuromuscular and adult onset disorders such as Alzheimer's disease). For some conditions (adult onset) the option of knowing if pathogenic variants are present is given.
- Uncertainty - We may not be able to tell you with certainty whether the variant(s) we find are directly related to the patient's phenotype. The interpretation of WES will evolve over time as we learn more about normal and abnormal human variation.
- Anxiety - Patients and family members may experience anxiety before, during, and/or after testing.

What will be reported?

- Any variants in genes that are considered to be related to phenotype will be reported. Please note that adult-onset conditions that are not currently medically-actionable will NOT be reported in individuals younger than18 years of age. These can be requested at a later date.

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Proband

The **ExomSure I Dx / MendeliomSure I Dx / PanelSure I Dx** is not designed to be a comprehensive test to identify carrier status or findings in adult-onset conditions. We are unable to guarantee that all conditions for which the individual is a carrier for or all adult conditions for which the individual has a pathogenic variant in will be determined by this test. Additional testing for health or reproductive purposes should be discussed with your doctor or genetic counselor. Also, variants of unknown significance will not be reported when they fall under these categories.

The risks, benefits, and limitation of **ExomSure I Dx / MendeliomSure I Dx / PanelSure I Dx** testing have been explained to me and I have had a chance to have my questions answered. I have read and will receive a copy of this consent form.

TRIOS

If any findings are identified in the requested optional disclosure categories in the proband, parental status for these specific findings can be released for parents undergoing the **ExomSure I Dx / MendeliomSure I Dx / PanelSure I Dx** as part of our Trios option. No separate report will be issued for the parents.

Please Note: The parental exome data is not analyzed for secondary findings and parents can carry other variants that are not inherited by the proband which willtherefore not be reported. Additional testing for health purposes should be discussed with your doctor or genetic counselor.

If information about the parental status is wanted, please initial next to the appropriate response and sign your name below.

Physician/ Counselor/ Clinician Statement

I have provided genetic counseling and have explained the ExomSure I Dx / MendeliomSure I Dx / PanelSure I Dx test to the patient/parent/guardian. The consent form and limitations of genetic testing were viewed with the patient/guardian. I accept responsibility for pre- and post-test genetic counseling.

Note to Ordering Clinician

Eurofins Clinical genetics India Pvt. Ltd. encourages the discussion of the limitations and utility of a genetic test with the patient prior to specimen collection. This form is provided to address pertinent issues regarding this test. Specific information describing indications, methodology and detection can be found on the Eurofins Clinical genetics India website at: *www.eurofinclinicalgenetics.co.in*

I can request that remaining sample can be used for research purposes. ☐ **Yes** ☐ **No**

		Signature	Date
Proband			
Trios	Mother / Guardian		
	Father / Guardian		
Additional Family Member			
Physician / Clinician			