Ambry Genetics®

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1. TEST PROCESS

ExomeNext-***Select*** involves sequencing of the ~20,000 nuclear genes and analysis of up to 500 genes pre-selected by the ordering clinician. This process may include analysis of genes that have been previously associated with human disease (characterized) as well as those that have not been previously described to cause a Mendelian condition (novel/uncharacterized). Whole exome sequencing differs from whole genome sequencing as it targets the ~1-2% of the protein coding regions (exons) of the genome. The goal of ExomeNext-*Select* is to identify the underlying molecular cause of an affected individual's condition.

2. TECHNICAL LIMITATIONS

Not all exons in the genome are targeted. Approximately 1-2% of the exons that are targeted may not be well covered. The empirical coverage data for specific genes can be found on the Ambry Genetics website. Certain mutation types may not be detectable (eg. some copy number variants, methylation abnormalities, mutations in genes with highly homologous pseudogenes, and expansions of trinucleotide repeats) and exome sequencing is also limited in the detection of alterations confounded by various non-Mendelian factors (penetrance, variable expressivity, multifactorial disease, epigenetic factors, phenocopies and uniparental disomy (UPD)).

3. TESTING & ANALYSIS PIPELINE

Several hundred thousand variants will be identified through whole exome sequencing, and all variants will be filtered through an in-house developed pipeline, Ambry variant analyzer (AVA), based on types of alterations, minor allele frequencies, and various mutation databases. Analysis will be limited to the genes selected by the ordering clinician. No other genes will be analyzed or reported.

4. TESTING OF FAMILY SAMPLES

Co-segregation analysis (family studies) is performed on family members submitted at the time of testing for potentially informative variants. Providing family member samples can improve the likelihood of a more definitive diagnosis.

Confirmation of reportable findings by Sanger sequencing will be performed for all alterations that fail to meet quality thresholds. De-identified co-segregation results for the family members will be included in the primary report. If no reportable findings are identified, family member samples will not be tested. Testing of family members submitted after ExomeNext-*Select* testing is completed is available at standard Specific Site Analysis pricing.

5. CLINICAL COURSE/PROGNOSIS OF DISEASE

Identification of a specific genetic variant does not predict the onset, severity, or spectrum of human disease with any degree of certainty. Similarly, the absence of a sequence variant may reduce, but will not eliminate the possibility of being affected with a specific condition.

6. STANDARD LABORATORY LIMITATIONS

Standard laboratory limitations apply to each specimen drawn for testing, including but not limited to: sample mix-up, samples unavailable from critical family members, inaccurate reporting of family relationships, mosaicism, low-level heteroplasmy or technical limitations. Under these potential, yet rare circumstances, exome sequencing may not be capable of generating an accurate result.

7. SECONDARY FINDINGS

Secondary findings are not analyzed or reported for ExomeNext-*Select* test orders.

8. TERMS AND CONDITIONS

Patient Acknowledgement: I acknowledge that the information provided by me is true and correct. For direct insurance billing: I authorize my insurance benefits to be paid directly to Ambry Genetics
Corporation (Ambry), authorize Ambry to release medical information concerning my testing to my insurer, to be my designated representative for purposes of appealing any denial of benefits as needed and to request additional medical records for this purpose. I understand that I am financially responsible for any amounts not covered by my insurer and responsible for sending Ambry money received from my health insurance company.

For patient payment by credit card: I hereby authorize Ambry Genetics Corporation to bill my credit card as indicated above. In order to expedite consideration for eligibility for Ambry's Patient Assistance Program, please provide the total annual gross household income: $ _____ and the number of family members in the household supported by the listed income: _____ .
I authorize Ambry Genetics Corporation to verify the above information for the sole purpose of assessing financial need, including the right to seek supporting documentation.

Initials

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☐ For NY Residents: By checking this box, I agree that Ambry Genetics will retain my sample for 6 months after the testing above has been completed. By not checking this box, I understand that under New York State law, Ambry Genetics must discard my sample after the longer of (a) testing completion and (b) 60 days after the Date of Collection above.

I have read or have had read to me all of the above statements and understand the information regarding molecular genetics testing and have had the opportunity to ask questions I might have about the testing, the procedure, the risks, and the alternatives prior to my informed consent. I agree to have the molecular genetic testing described within or above.

Patient Signature *(or Parent/Guardian if patient is a minor)*

Date

Patient Name *(Print)*

Name and Relationship *(Parent/Guardian if patient is a minor)*

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