Amby Lab

Clinical Exome analysis service

Amby Lab is making high-quality genetic testing for rare hereditary disorders applying next-generation sequencing to identify alterations in human Clinical Exome.
Exome sequencing allows to identify all four type of alterations across the genes of interest. The test simultaneously sequences the coding regions of 5087 clinicaly-relevant genes and 268 SNPs. The complete list of genes that are testes appears in the “Gene List” table.

Amby Lab Exome sequencing detects base substitutions, insertions and deletions (InDels), copy number variations (CNV) and rearrangements using genomic DNA, obtained from any material (i.e. saliva, blood etc.).

The following pipeline demonstrates the overview of our service:

Starting material	• 1-2 mkg of human Genomic DNA • DNA should be tested for degradation and RNA contamination
Laboratory processing	• Starting material QC • Library preparation • Exome enrichment (12 Mb panel)
NGS	• High-throughput sequencing on Illumina HiSeq 2500 platform • 100 base pair-end reads, 1.5 Gb of data, x70 mean coverage • Coverage of Clinical exome panel by reads: >80% • Effectiveness of enrichment (amount of reads mapped to the panel, rather than to other genome positions): >50%
Output	• Raw sequencing data in FastQ format
Additional services	• Bioinformatic analysis of exome sequencing data: read mapping and variant calling. Outputs are BAM and VCF files • Interpretation of VCF file using Amby Lab online medical system • Interpretation of results by certified Medical geneticist using clinical records

We offer three levels of the service:

SERVICE LEVELS			
PROCEDURE	**LEVEL 1**	**LEVEL 2**	**LEVEL 3**
1. Initial sample QC	●	●	●
2. Library preparation	●	●	●
3. Exome enrichment	●	●	●
4. NGS	●	●	●
5. Bioinformatic analysis		●	●
6. Processing of data by medical system with interpretation of results by medical geneticist*			●

NOTE

* Medical interpretation requires clinical records, provided by the customer

DELIVERABLES			
OUTPUT DATA	**LEVEL 1**	**LEVEL 2**	**LEVEL 3**
1. FastQ file (raw data)	●	●	●
2. BAM and VCF files		●	●
3. Exome analysis report			●

Service levels may be adjusted for your particular needs

Extra option:

Amby Lab online Medical interpretation system allows to filter out clinically irrelevant mutations (not associated to inherited diseases) found during Exome sequencing. The interpretation could be made by the customer as well. The input data for interpretation is VCF file. All found mutations are processed and characterized for relevance to inherited diseases.
User can use a number of the following build-in filters:

- Disease database filter - filter altered genes associated with diseases according to individual's clinical symptoms. Support OMIM database naming. The filter allows to select all genes associated to the disease or subset some portion of them.
- Disease panel filter – filter genes with mutations associated to commonly used Disease panels
- Single gene filter – allows to add particular gene to a list

Amby Lab online Medical System is available at: medsys.ambylab.com