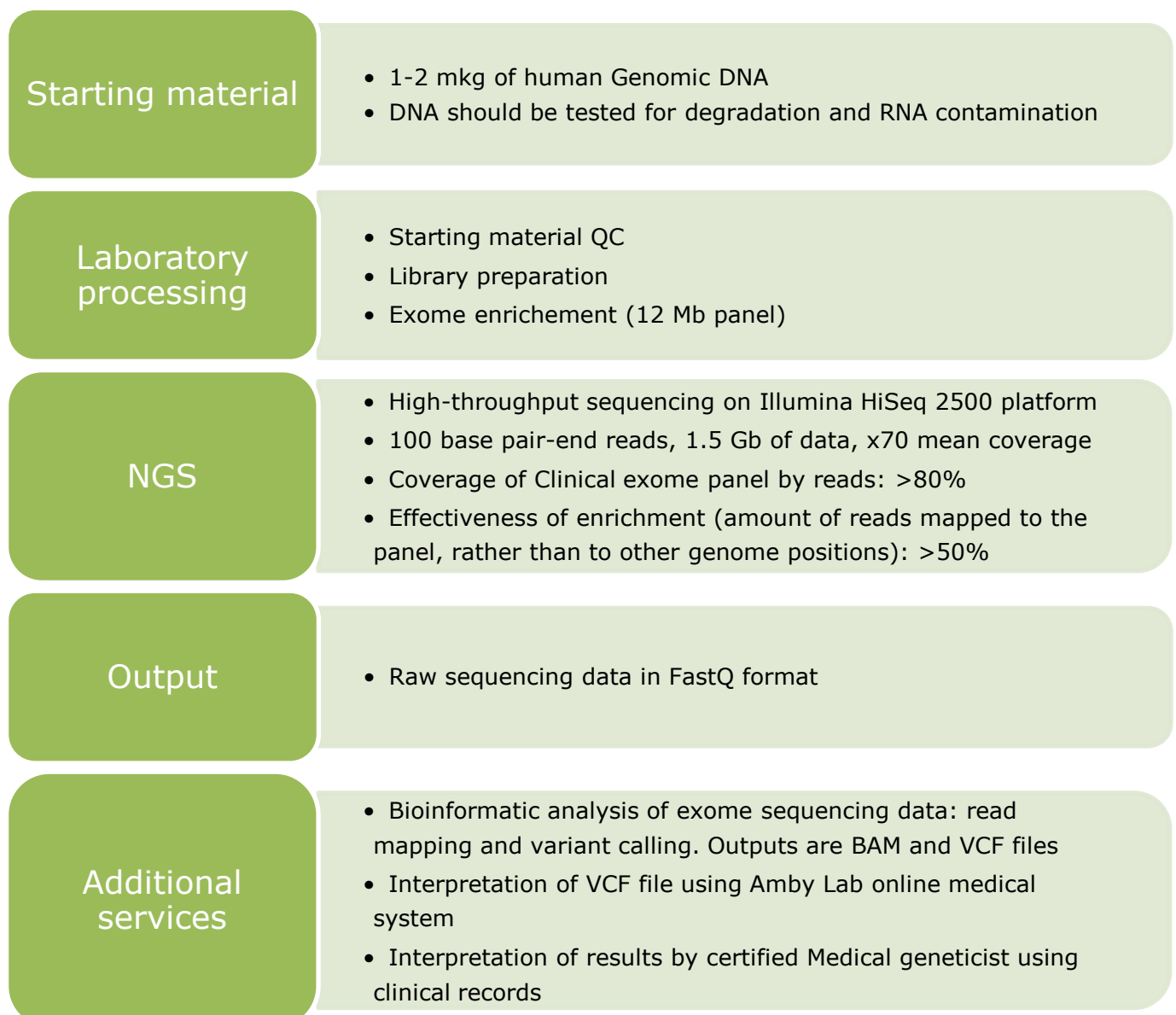


## Clinical Exome sequencing service

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 clinically-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.



Amby Lab online Medical interpretation system allows to filter out clinically irrelevant mutations found during Exome sequencing. The input data for interpretation is VCF file. All found mutations are processed and characterized for relevance.

User can use a number of the following build-in filters:

- Diseases 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](https://medsys.ambylab.com)