Research &
Pharma Solutions

ExomeXtra® Sequencing

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With the launch of our new ExomeXtra® Sequencing (EXS) products, we launched a whole new service category in the field of Research and Pharma Solutions (RPS). The ExomeXtra® Sequencing service category is located between the service categories Whole Genome Sequencing (WGS) and Whole Exome Sequencing (WES), as it closes the gap between genome and exome sequencing.

As in exome sequencing, all known coding exons are enriched in our ExomeXtra® Sequencing products. They are enriched and sequenced with a sufficient sequencing depth for confidentially calling variants at specific locations. However, additional genetic information might be required to solve complex disease patterns. For this purpose, our ExomeXtra® Sequencing products also include disease-relevant variants located in non-coding, intronic, or regulatory regions. With this extension, genome regions described as disease-relevant in respective databases are covered by ExomeXtra® Sequencing.

To enable genome-wide analyses and high-resolution detection of copy number variations (CNVs), we included a new and unique CNV backbone into our ExomeXtra® enrichment. With this CNV backbone, deletions and duplications in the entire genome can be detected, increasing the probability of finding disease-causing mutations. The CNV backbone is closing the gap to genome sequencing.

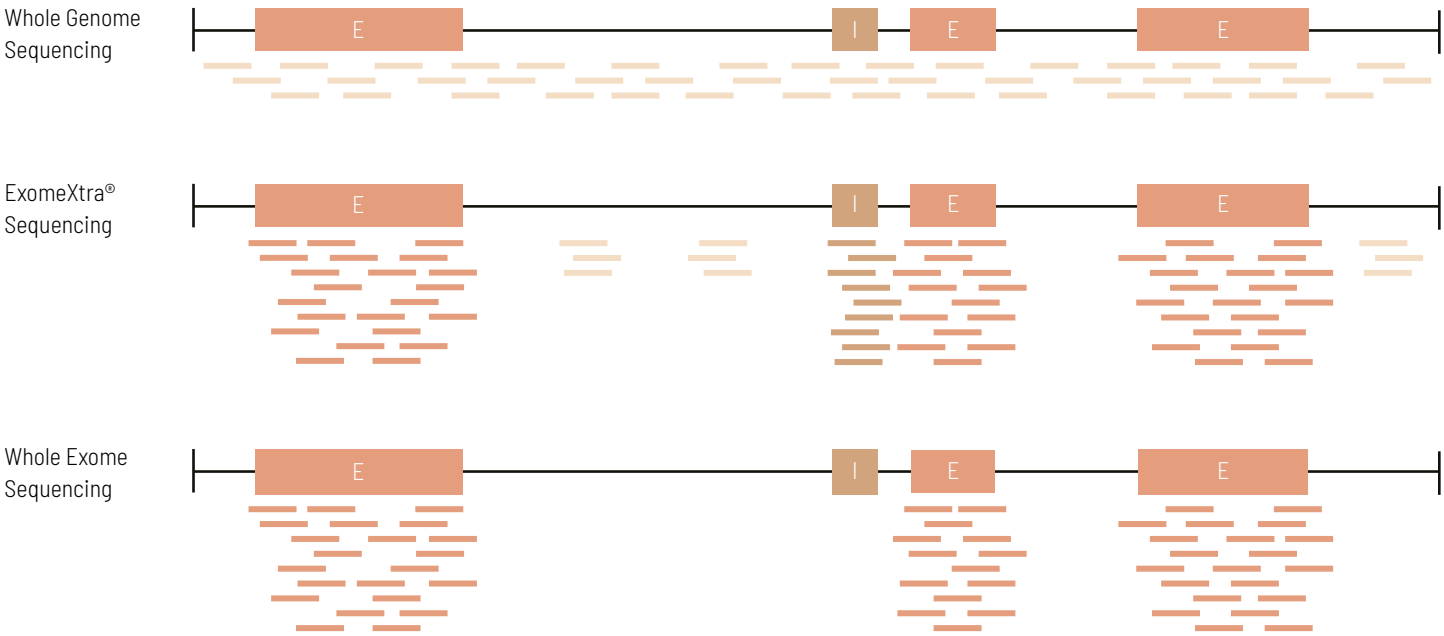

Figure 1: ExomeXtra® Sequencing closes the gap between Whole Genome Sequencing and Whole Exome Sequencing. In Whole Genome Sequencing (WGS), the whole genome is sequenced, independent of exons (E) or disease-relevant intronic variants (I). In Whole Exome Sequencing (WES), exonic regions are sequenced with a sufficient sequencing depth to confidentially call variants. ExomeXtra® Sequencing closes the gap between genome and exome sequencing by using a CNV backbone for genome-wide CNV calling, inspired by WGS, and enriching all disease-relevant variants in coding and non-coding regions to confidentially identify the genetic cause of a disease.

ExomeXtra® Sequencing combines the advantage of exome sequencing with the advantages of genome sequencing. The output is significantly smaller for ExomeXtra® Sequencing than for our Whole Genome Sequencing, but the additional features require a higher output than for our classical Whole Exome Sequencing products (see table 1).

To allow a constant improvement of the analysis of disease-relevant targets in coding and non-coding regions according to the latest scientific knowledge, regular updates of our ExomeXtra® Sequencing products are required. During these updates, new non-coding variants from relevant databases, such as HGMD or ClinVar, that are classified as pathogenic or likely pathogenic are included in our ExomeXtra® enrichment. These regular updates are a crucial element of our ExomeXtra® Sequencing products. As we always want to provide the latest scientific knowledge, we only offer the latest version of our ExomeXtra® enrichment.

Table 1: Comparison of Whole Genome Sequencing (WGS), ExomeXtra® Sequencing (EXS), and Whole Exome Sequencing (WES).

	WGS Large Classic	EXS Classic	WES Classic
Targeted region	Whole genome	Whole exomic region + mitochondrial genome + relevant non-coding regions + CNV backbone	Whole exomic region + mitochondrial genome
Output	90 Gb	18 Gb	12 Gb

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