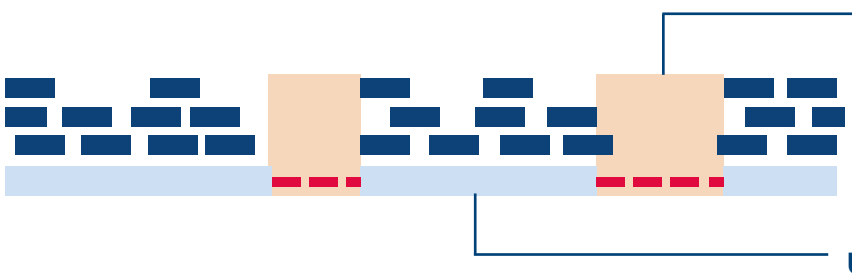
We've got you covered

With the **QIAseq® Human Exome** solution for rare variant detection

>60% of disease-causing mutations are found in protein-coding regions of the genome.

Compared to whole genome sequencing (WGS), **whole exome sequencing (WES)** focuses on coding regions, offering powerful variant detection power while **reducing time and expense**.

Uniform sequencing coverage is crucial

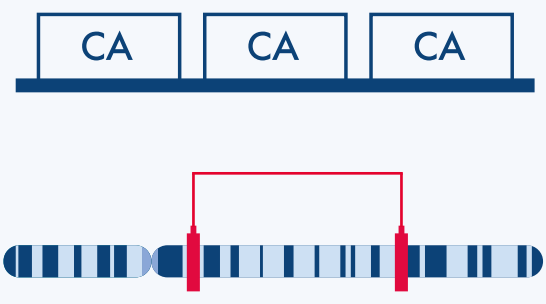


Uneven coverage can result in low coverage of important genomic regions.

Uniform coverage is required for efficient variant discovery.

Causes of uneven coverage

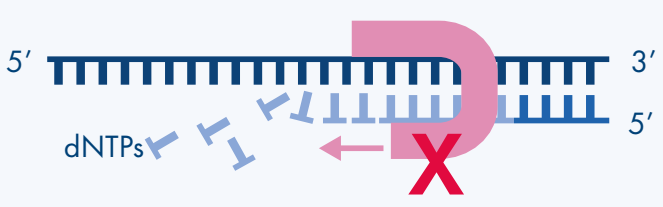
Uneven sequence coverage is linked to **repeat elements** and **segmental duplications**.



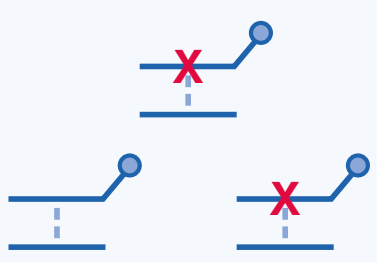
But is mainly attributed to **>65% GC content**

High GC content reduces the efficiency of:

01 PCR amplification



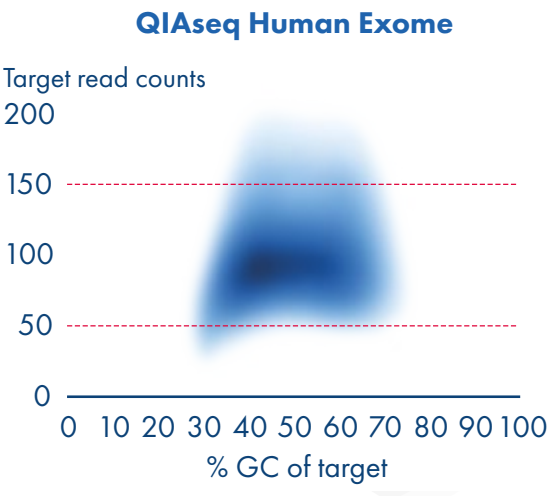
02 Capture probe hybridization

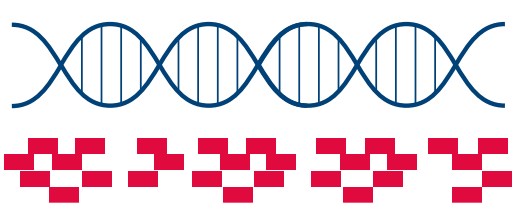


Overcoming GC bias

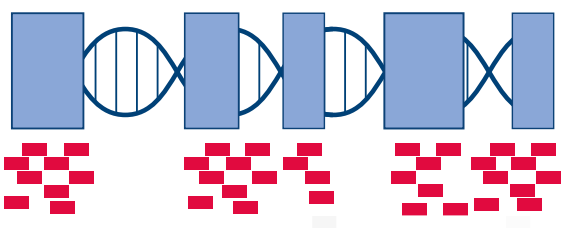
The **QIAseq Human Exome** solution is optimized to **minimize GC bias**, giving uniform exome coverage.

Plus, **QIAseq Human Exome** provides many reads per position, delivering WGS-quality variant data without the expense.



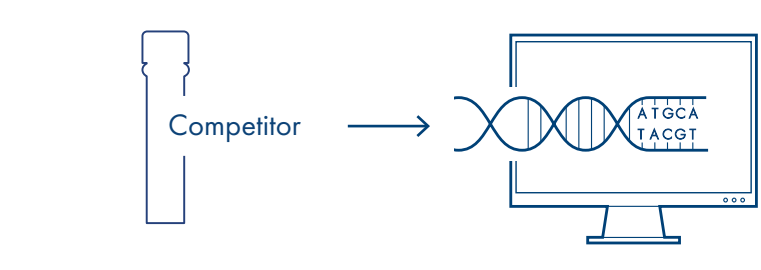


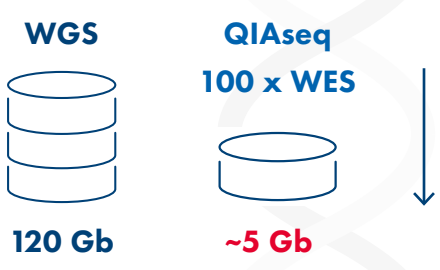
WGS

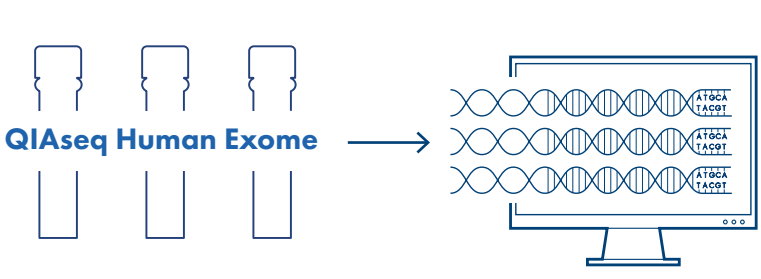


WES with QIAseq

QIAseq Human Exome reduces sequencing needs by **50%**, allowing more samples per run but produces just **~5 Gb** of data.







As a result, QIAseq Human Exome reduces costs by **30–50%**.

QIAseq Human Exome was built with researchers' needs in mind

6 sample types supported: gDNA derived from whole blood, cells, tissue, saliva, FFPE and cfDNA

1-day workflow, compatible with automation and scalable

50% reduction in sequencing costs

33% less time than competitors

References

1. Wang, Q. et al. (2017) Novel metrics to measure coverage in whole exome sequencing datasets reveal local and global non-uniformity. Sci. Rep. 7, 885.
2. Chilamakuri, C.S., et al. (2014) Performance comparison of four exome capture systems for deep sequencing. BMC Genomics. 15, 449.

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