Olink®

Proteomics glossary

Protein quantitative trait loci (pQTLs)

DNA variants that affect protein levels in blood or other tissues, they:

- Link genetic variation to protein expression—connecting genome to phenotype traits
- Help uncover disease mechanisms and accelerate therapeutic development

Olink's NGS-based proteomics has powered major studies such as the UK Biobank Pharma Proteomics Project, enabling the discovery of over 14,000 genetic associations with protein expression levels.

Cis-pQTLs

Genetic variants located near (<1 Mb) the gene encoding a specific protein, they:

- Influence the expression levels of that protein
- Provide insights into the genetic regulation of proteins
- Help determine whether a protein plays a causal role in disease

Olink's NGS-based proteomics platform, with its exceptional specificity, enables:

- Robust identification of cis-pQTLs
- Prioritization of SNPs
- Deeper understanding of disease mechanisms and causality

Trans-pQTLs

Genetic variants that affect proteins encoded by genes on different chromosomes or distant loci, they:

- Reveal novel regulatory pathways
- Provide insights into biological mechanisms underlying disease

Large-scale efforts (e.g., the SCALLOP consortium) using Olink's NGS-based proteomics have:

- Mapped novel trans-pQTLs across large cohorts
- Advanced understanding of trans-regulation
- Exposed molecular connections in human biology

Mendelian randomization (MR)

A robust method in genetics and epidemiology to:

- Assess whether an observed association between a risk factor (e.g. protein levels) and a disease is likely to be causal.
- In pQTL studies to test whether changes in protein levels (influenced by cis- or trans-pQTLs) actually cause disease, not just correlative.

Combined with Olink's high-quality proteomics data, MR enables:

- Trusted strategy for identification of causal proteins
- Prioritization of drug targets and pathways
- Acceleration of precision medicine

Polygenic risk score (PRS)

Estimates an individual's genetic predisposition to a trait or disease, it:

- Is calculated by aggregating the effects of many genetic variants across the genome, each variant contributes a small effect to overall risk
- Provides insight into how genetic makeup influences the likelihood of developing specific conditions

Proteomic risk score (ProRS)

Predicts an individual's disease risk based on the combined analysis of multiple protein biomarkers in biological samples (e.g. plasma).

Using NGS-based proteomics technologies like Olink® Explore and Olink® Reveal, ProRS enables:

- Identification and quantification of proteins associated with specific disease states.
- Baluable insights into disease mechanisms, helps uncover potential therapeutic targets,
- More accurate risk prediction.

Olink NGS-based proteomics

Combines antibody-based assays with next-generation sequencing (NGS) for protein identification and quantification

- Uses DNA barcodes and NGS sequencing (not mass spectrometry) to detect proteins
- Delivers high specificity, sensitivity, and scalability
- Ideal for researchers with existing NGS workflows—even those new to proteomics.

Proximity extension assay (PEA™)

Combines antibody-based assays with next-generation sequencing (NGS) for protein identification and quantification

- Uses DNA barcodes and NGS sequencing (not mass spectrometry) to detect proteins
- Delivers high specificity, sensitivity, and scalability
- Ideal for researchers with existing NGS workflows—even those new to proteomics

NPX (normalized protein expression)

Olink's unit for relative protein abundance

- Derived from Olink's NGS-based proteomics platform measurements
- Logarithmically related to protein concentration
- Normalized to reduce technical variation—enabling reliable comparisons across samples and studies

NPX has become a de facto standard in proteomics measurement

- Widely adopted in large cohort studies such as the UK Biobank-PPP
- Enables researchers to:
    - Benchmark data against large reference datasets
    - Accelerate cross-study integration in multiomics research

1633, v1.0, 2025-15-05

olink.com/ngs-based-proteomics