



# Release Notes

Release Date: Dec 3rd, 2025

## New Features

- Activity Scores on Phenotype Table for CYP2D6, CYP2C9, and DPYD
- Unreported Allele Insights
- Analytics Risk Stratification Custom Query Builder
- SmartStart/Onboarding Dashboard

## Enhancements

- Create patient profiles in CDSS with JSON upload
- Software Update Version in status report
- Download ULR format from LabGx
- Acceptance of VCF files
- Full .NET 10 upgrade

## New Features

### Activity Scores on Phenotype Table for CYP2D6, CYP2C9, and DPYD:

A new column has been added to your Phenotype Summary Table that displays the Activity Score for CYP2C9, CYP2D6, and DPYD genes. **For these genes, CPIC uses an “activity value” (AV) to describe a graded activity for alleles. When the activity values of the maternal and paternal alleles are summed, the resulting “activity score” (AS) assists in assigning phenotype.**

Phenotype Table

| Gene    | Genotype Result | Activity Score | Phenotype Result OR Genotype Explanation |
|---------|-----------------|----------------|------------------------------------------|
| CYP2B6  | *1/*1           | N/A            | Normal Metabolizer                       |
| CYP2C19 | *38/*38         | N/A            | Normal Metabolizer                       |
| CYP2C9  | *1/*2           | 1.5            | Intermediate Metabolizer                 |
| CYP2D6  | *1/*9           | 1.25           | Normal Metabolizer                       |
| CYP3A4  | *1/*1           | N/A            | Normal Metabolizer                       |
| CYP3A5  | *3/*3           | N/A            | Poor Metabolizer                         |
| DPYD    | *1/HapB3        | 1.5            | Intermediate Metabolizer                 |
| NUDT15  | *1/*1           | N/A            | Normal Metabolizer                       |
| SLCO1B1 | *1/*1           | N/A            | Normal Function                          |
| TPMT    | *1/*1           | N/A            | Normal Metabolizer                       |

Activity Score plays a key role in drug-gene recommendations where the degree of reduced function affects management. In this release, guidance for DPYD-related recommendations involving capecitabine and fluorouracil has been expanded.

Previously, messaging was simplified; now your LabGx reports include **full CPIC-aligned details**.

For individuals with a DPYD Poor Metabolizer phenotype, CPIC generally recommends avoiding capecitabine and fluorouracil. When the Activity Score is 0.5 (rather than 0), CPIC provides additional considerations. **Clinicians should confirm the patient’s Activity Score in the phenotype table before applying these recommendations**, and specialist involvement is strongly advised when managing these high-risk therapies.

You can enable this setting within Lab Studio - Technical Report settings under the option Include Activity Score. This is turned on by default for new reports to support more precise recommendations for medications such as those impacted by DPYD, but you can disable it in your PDF template settings if preferred.

For genes with copy number values such as CYP2D6, all possible Activity Scores based on allele duplications will be displayed within your phenotype table to give you a comprehensive view:

|        |            |               |
|--------|------------|---------------|
| CYP2D6 | (*4/*10)4N | 0.75,0.5,0.25 |
|--------|------------|---------------|

| fluorouracil                           | Phenotype                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            | Genetic Test | Results                     | Source/Evidence Level                     |
|----------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------|-----------------------------|-------------------------------------------|
| Carac<br>Efudex<br>Fluoroplex<br>Tolak | Poor metabolizer                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | DPYD         | HapB3/c.295_298delTCAT (*7) | CPIC A <sup>4</sup> ; FDA 1 <sup>46</sup> |
| ReviewGx                               | <b>Drug-Gene Interactions:</b><br><b>Serious drug-gene interaction: avoid/select alternative</b><br><br>⚠ CPIC – Implication: Complete DPD deficiency and increased risk for severe or even fatal drug toxicity when treated with fluoropyrimidine drugs.<br><br>⚠ CPIC – Strong Recommendation: Avoid use of 5-fluorouracil or 5-fluorouracil prodrug-based regimens.<br><br><b>For Activity Score 0.5 only (refer to phenotype table for patient’s activity score result):</b> In the event, based on clinical advice, alternative agents are not considered a suitable therapeutic option, 5-fluorouracil should be administered at a strongly reduced dose with early therapeutic drug monitoring.<br>If available, a phenotyping test that assesses DPD enzyme activity should be considered to estimate the starting dose. In absence of phenotyping data, a dose of <25% of the normal starting dose is estimated assuming additive effects of alleles on 5-FU clearance. Therapeutic drug monitoring should be done at the earliest time point possible (e.g., minimum time point in steady state) in order to immediately discontinue therapy if the drug level is too high.<br><br><b>Drug-Drug Interactions:</b><br>⚠ (Leucovorin)-(Fluorouracil) The risk or severity of adverse effects can be increased when Leucovorin is combined with Fluorouracil. |              |                             |                                           |

#### Technical Report

- ☐ Include Genomic Locations
- ☒ Include Phenotype Definitions
- ☒ Include Activity Scores

#### Please note:

This feature is now turned on by default for everyone

## New Features

### Unreported Allele Insights

You now have access to Unreported Allele Insights in your lab director override window to help you manage reports with unreported alleles caused by common genotyping data inconsistencies. When you enter the override window and press the Annotate button, you will see a new Error column displaying the possible reasons for unreported alleles. By clicking the circular warning icon next to an error explanation, you can open the error message window.

Within this window, you'll find two tabs: Testing Location, showing all testing locations linked to the gene; and Copy Number, displaying the copy number values associated with the sample. The testing locations responsible for the unreported alleles are highlighted and bolded in the table to help you quickly identify the source of the issue.

This feature, designed specifically for lab directors or others completing report sign-off, will streamline your review process by clearly pointing out testing locations which caused the unreported alleles. It addresses three key scenarios causing unreported results: unreported genotypes, heterozygous results when a CNV of 1 is reported for CYP2D6, and genotype results when a CNV of 0 is present for CYP2D6.

The screenshot illustrates the 'Unreported Allele Insights' feature in the GenXys LabGx interface. The main window shows an 'Overrides' table with columns: Gene, Type, Value, Omit, Modified, and Error. The 'Error' column displays 'Unreported/Unknown genotype (SNP)' with a warning icon. A red arrow points from the 'Annotate' button in the top right to the warning icon. Another red arrow points from the warning icon to a pop-up window titled 'Unreported/Unknown genotype (SNP)'. This pop-up window has two tabs: 'Testing Location (21)' and 'Copy Number (1)'. The 'Testing Location' tab is active, showing a table with columns: Gene, RSID, Converted Genotype, and Unconverted Genotype. The table lists several CYP2D6 variants, with the row for RS16947 (UK/UND) highlighted. Below the table, it says 'Most likely due to poor QC/DNA quality'. There are 'Clear' and 'Generate' buttons on the right, and a 'Close' button at the bottom right of the pop-up.

| Gene   | Type      | Value | Omit | Modified | Error                             |
|--------|-----------|-------|------|----------|-----------------------------------|
| CYP2D6 | Genotype  | UK    | No   |          | Unreported/Unknown genotype (SNP) |
| CYP2D6 | Phenotype | UK    | No   |          |                                   |

| Gene          | RSID           | Converted Genotype | Unconverted Genotype |
|---------------|----------------|--------------------|----------------------|
| CYP2D6        | RS1065852      | G/G                | G/G                  |
| CYP2D6        | RS1135822      | A/A                | A/A                  |
| CYP2D6        | RS1135840      | C/G                | C/G                  |
| <b>CYP2D6</b> | <b>RS16947</b> | <b>UK</b>          | <b>UND</b>           |
| CYP2D6        | RS201377835    | C/C                | C/C                  |
| CYP2D6        | RS267608319    | C/C                | C/C                  |

Most likely due to poor QC/DNA quality

For more details, refer to the full information here [👉](#)

## New Features

### Analytics Risk Stratification Custom Query Builder

You can now use the Custom Query Builder within the Analytics Risk Stratification module (formerly known as IdentifyGx) to create tailored queries based on your specific criteria, enabling deeper insights into patient risk profiles and population-level trends.

#### Key features include:

- Multiple Queries: Build several queries at once, each generating independent results.
- Flexible Logic: Group conditions with AND/OR operators, including nested combinations.
- Custom Conditions: Define categories, operators, and thresholds to precisely match your needs.
- Visual & Intuitive: Color-coded logic (AND in green, OR in orange) and simple expand/collapse controls.
- Easy Management: Add, delete, or reset queries at any time.

This tool empowers you to generate precise, actionable risk analytics. For detailed instructions, refer to the [Analytics Dashboard User Manual. \(link\)](#)

RISK STRATIFICATION

Hide ☐

Query Builder

13 queries ^

Expand All ☐

RESET

Query #13 Drug-Gene Interactions (DGI) (>0) and (Cardiovascular Condition (>0) or Mental Health Condition (>0) or Drugs (>4))

Group Operator: AND

Category

Drug-Gene Interactions (DGI)

Op

>

Threshold

0

+

-

AND

Group Operator: OR

Category

Cardiovascular Condition

Op

>

Threshold

0

+

-

OR

Category

Mental Health Condition

Op

>

Threshold

0

+

-

OR

Category

Drugs

Op

>

Threshold

4

+

-

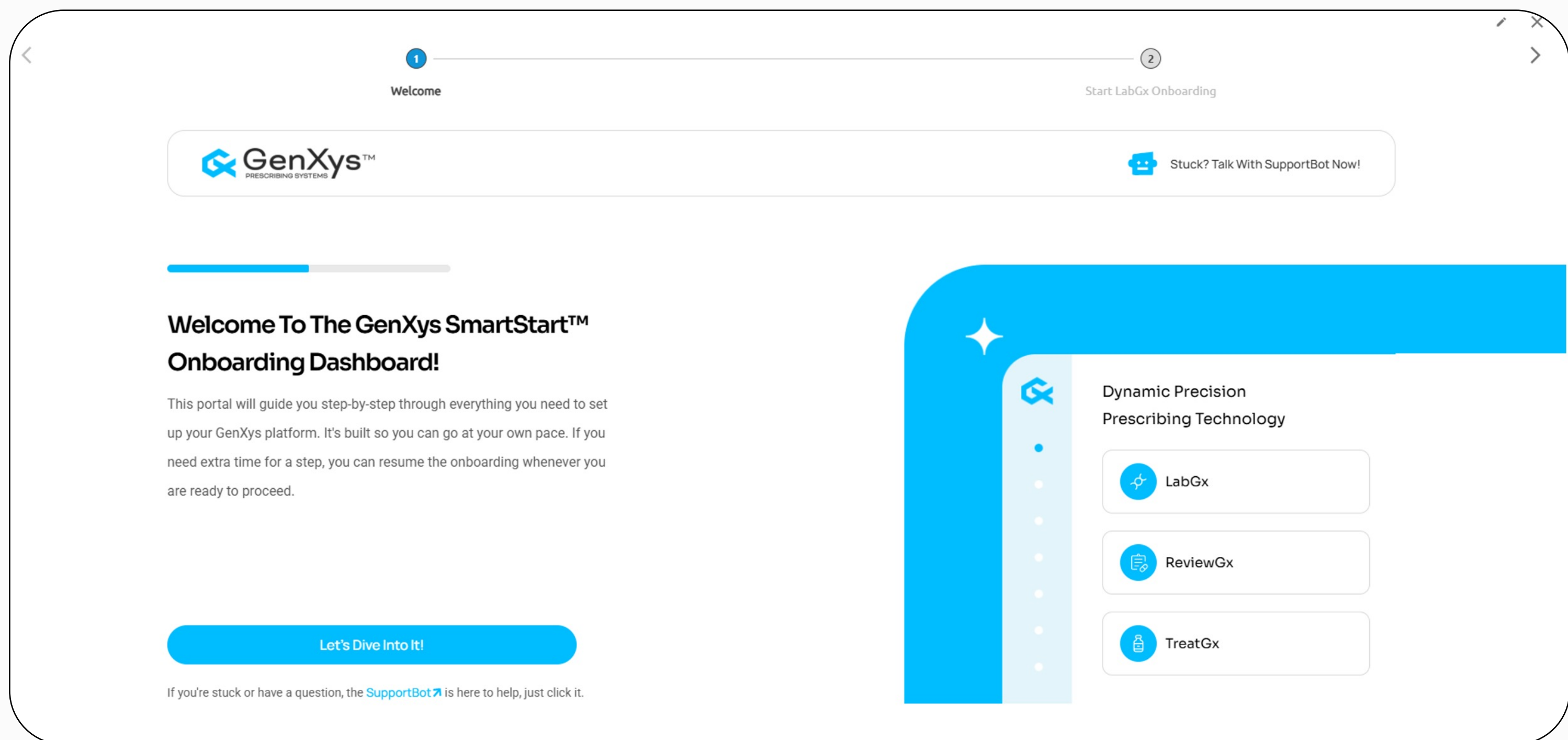
+ ADD QUERY

## New Features

### SmartStart/Onboarding Dashboard

You can now access the SmartStart Onboarding Dashboard, a centralized self-serve hub designed to streamline your lab's setup process, complete onboarding tasks, access training resources, and manage your LabStudio content.

Through this dashboard, conveniently located in the top-middle section of your lab portal page (open it using the arrow button), you can adjust mappings and explore new features.



To learn more, here is a quick overview video: [GenXys Smart Start](#) 

## Enhancements

# Create patient profiles in Clinical Decision Support Software (CDSS) with JSON upload

You can now create patient profiles in the CDSS (TreatGx and ReviewGx Portal) more quickly and efficiently by uploading a JSON file containing demographic and medication information, eliminating manual entry for each field. This uses the same JSON schema as order uploads for consistency and standardization, saving you time and reducing errors during patient setup.

How to Upload a New Patient:

1. Open ReviewGx or TreatGx.
2. From the left-hand menu, click Add Patient.
3. On the patient profile page, drag and drop your JSON file.
4. Once uploaded, click Save to create the profile.

For reference, the JSON order specification can be found here: [GenXys Lab Order Specification](#).



## Patient Profile

Expand All ☐

Order Pharmacogenetic Test

Last ordered: N/A

Order state:

 View Pharmacogenetic PDF

Drag and Drop Patient



## Enhancements

### Software Update Version in status report

The Status Report now includes an enhancement: Software Version Identification for PGxService (the LabGx software version). Each status report will display the software version that was used when the report was generated. This version number updates automatically with every platform improvement or update, providing you with clear traceability and helping you track which software iteration produced each report.

#### Status Report

|                         |                      |
|-------------------------|----------------------|
| <b>Report Generated</b> | 27/Oct/2025          |
| <b>Software Version</b> | PGxService: v7.0.219 |
| <b>Template</b>         | Default              |

## Enhancements

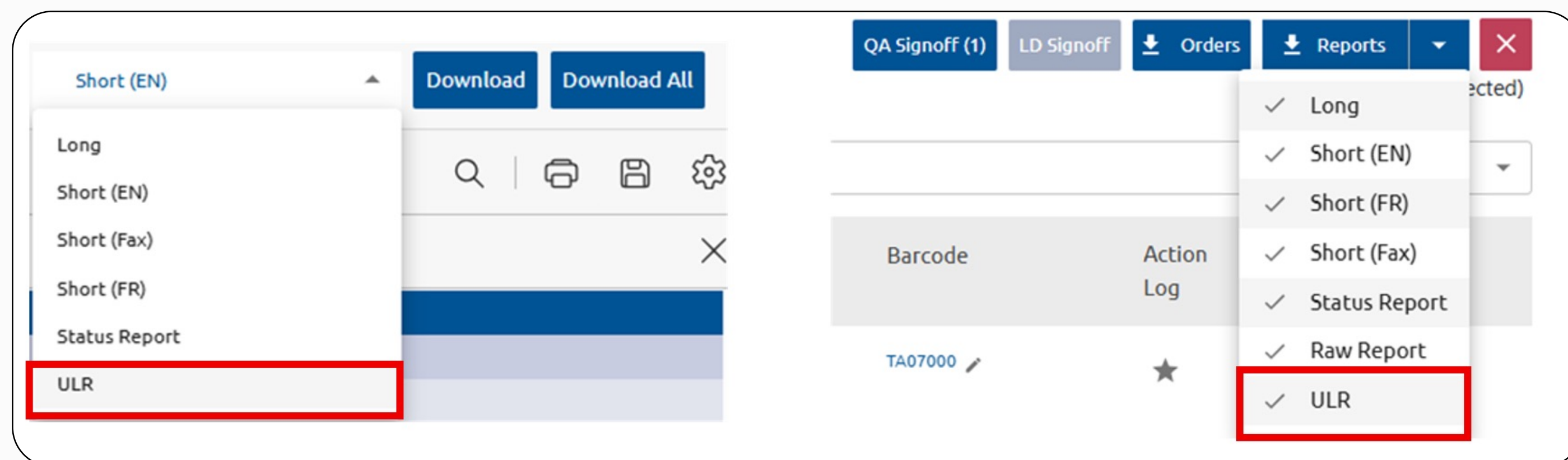
### Download ULR format from LabGx

You can now download ULR (Unified Lab Result) files for samples directly through the LabGx download function. These JSON-formatted files, generated internally from imported sample data, are available via batch download, individual file download, or API connection.

This enhancement provides you with easy access to standardized lab result data for further analysis or integration.

For detailed specifications, please visit

<https://www.genxys.com/wp-content/uploads/ULR-File-Specification-1.pdf> 



Enhancements

Acceptance of VCF files

LabGx now supports uploading VCF and gVCF files as sample files, making the platform ready for Next-Generation Sequencing (NGS) data. You can upload a single file per sample through both the API and web portal using the same process as before. Please ensure your files comply with at least the Variant Call Format (VCF) version 4.2 specification for compatibility. This update streamlines incorporating sequencing data into your workflows.

Drag & Drop Files

20A-253D0001.vcf

| Copy Number (.cnv) | Open Array (.oa) | HLA (.hla) | Further (.further) | Aux (.aux) | Mapping (.csv) | SNP (.snp) | STR (.str) | Unified Lab Result (.ulr) | Variant Call Format (.vcf or .gvcf) |
|--------------------|------------------|------------|--------------------|------------|----------------|------------|------------|---------------------------|-------------------------------------|
| ✕                  | ✕                | ✕          | ✕                  | ✕          | ✕              | ✕          | ✕          | ✕                         | ✓                                   |
| Property           |                  | Value      |                    |            |                |            |            |                           |                                     |
| Patients           |                  | None       |                    |            |                |            |            |                           |                                     |
| Details            |                  | None       |                    |            |                |            |            |                           |                                     |

Clear

Verify Files

Generate Reports

## Enhancements

### Full .NET 10 upgrade

To leverage the latest frameworks to improve performance, security, and long-term support, we've upgraded to .NET 10. This upgrade provides a more scalable and maintainable foundation for future feature development and integrations.



If you have any questions, feedback, or concerns,  
please contact [info@genxys.com](mailto:info@genxys.com) 🖱️