SOPHiA GENETICS
ZA La Pièce 12
CH – 1180 Rolle
Phone : + 41 21 694 10 60
VAT n° / TVA n° : CHE-184.818.745
sophiagenetics.com

Important Update: Variant Identifier harmonization

What you need to know about the Common Variant Identifier clean-up

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1. Overview

As part of our ongoing commitment to delivering accurate and consistent genomic insights, we have completed a harmonization process for variant identifiers across our platform. This document explains what changed, why we made this update, and what you may observe in your SOPHiA DDM™ environment going forward.

2. Background: Why we made this enhancement

When we migrated variant data from SOPHiA DDM™ (accessible through the desktop) to the New Generation of SOPHiA DDM™, we introduced a Common Variant Identifier (referred to internally as the CSGID) that allows our system to recognize a single variant across its different molecular representations (for example, trimmed, aligned, or lifted across reference genomes).

In SOPHiA DDM™ (accessible through the desktop), manual annotations, such as pathogenicity flags, variant descriptions, and false positive flags, were linked to each individual variant representation separately. During migration to the New Generation SOPHiA DDM™, variants with multiple representations could therefore carry more than one set of user annotations linked to the same underlying CSGID.

This variant normalization process brings meaningful improvements: it enables more accurate variant statistics and population frequencies, both within your account and across the broader user community.

3. What will change after Release v7.17.0

3.1 Conflict Resolution Logic

In rare cases, the same variant existed in multiple representations in SOPHiA DDM™ (accessible through the desktop), each carrying a different user annotation. To resolve these conflicts in a predictable and transparent way, we applied the following rule:

Resolution Principle

Where the same variant had conflicting user annotations across different representations, the annotation with the most recent modification timestamp was selected as the authoritative record in the New Generation of SOPHiA DDM™.

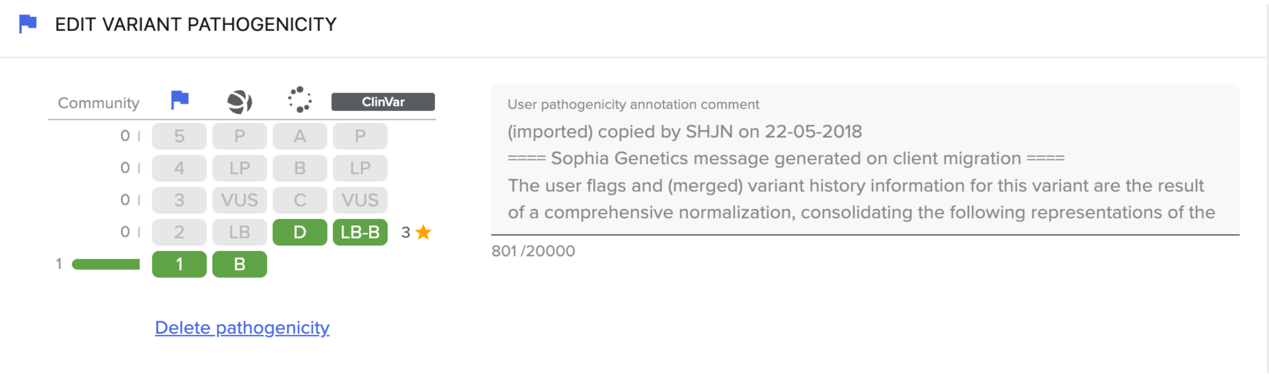
This one-time clean-up was applied to the following annotation types:

- User Pathogenicity (flag level and associated comment)
- Variant Description
- False Positive flag (flag level and associated comment)

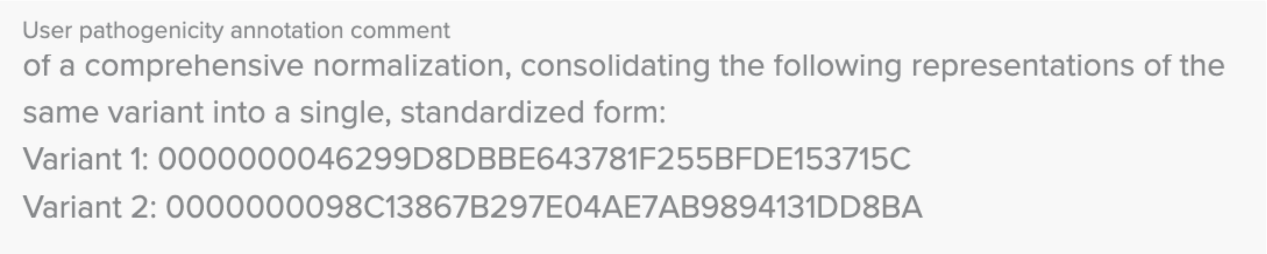
3.2 What you will see in the New Generation SOPHiA DDM™

After the clean-up:

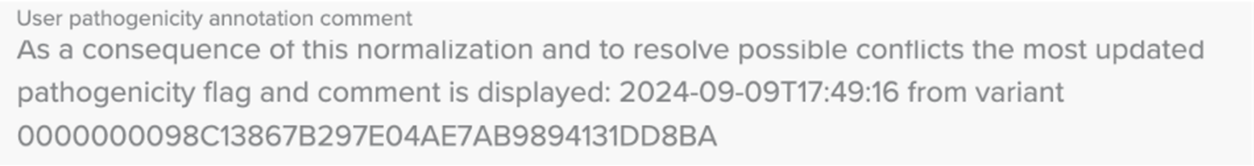
- The selected variant annotation is now shown in SOPHiA DDM™, with a system-generated comment appended to your existing user comment. This comment briefly explains the consolidation and lists the variant representation IDs that were involved.



User pathogenicity annotation comment
(imported) copied by SHJN on 22-05-2018
==== Sophia Genetics message generated on client migration ====
The user flags and (merged) variant history information for this variant are the result of a comprehensive normalization, consolidating the following representations of the

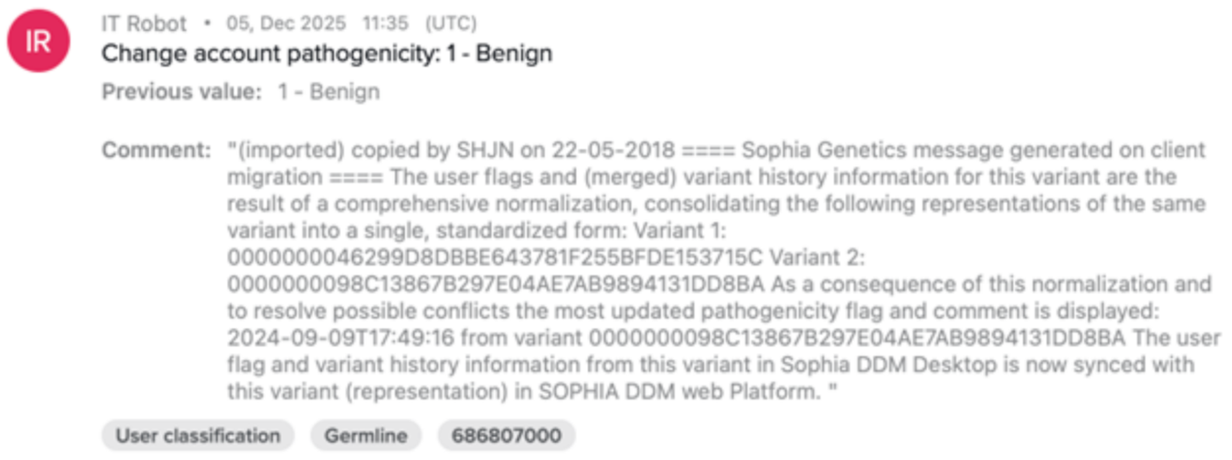


User pathogenicity annotation comment
of a comprehensive normalization, consolidating the following representations of the same variant into a single, standardized form:
Variant 1: 000000046299D8DBBE643781F255BFDE153715C
Variant 2: 0000000098C13867B297E04AE7AB9894131DD8BA



User pathogenicity annotation comment
As a consequence of this normalization and to resolve possible conflicts the most updated pathogenicity flag and comment is displayed: 2024-09-09T17:49:16 from variant 0000000098C13867B297E04AE7AB9894131DD8BA

- The full annotation history, including the activity from this clean-up process, is visible in the Variant History section, so the complete trail of user evidence is preserved.



IT Robot • 05, Dec 2025 11:35 (UTC)

Change account pathogenicity: 1 - Benign

Previous value: 1 - Benign

Comment: "(imported) copied by SHJN on 22-05-2018 ==== Sophia Genetics message generated on client migration ==== The user flags and (merged) variant history information for this variant are the result of a comprehensive normalization, consolidating the following representations of the same variant into a single, standardized form: Variant 1: 0000000046299D8DBBE643781F255BFDE153715C Variant 2: 0000000098C13867B297E04AE7AB9894131DD8BA As a consequence of this normalization and to resolve possible conflicts the most updated pathogenicity flag and comment is displayed: 2024-09-09T17:49:16 from variant 0000000098C13867B297E04AE7AB9894131DD8BA The user flag and variant history information from this variant in Sophia DDM Desktop is now synced with this variant (representation) in SOPHIA DDM web Platform. "

- History records from all representations of the same variant are always displayed together.

In SOPHiA DDM™ (accessible through the desktop), variant representations remain unchanged and independent. Each representation retains its individual user annotations as before.

3.3 Worked example

Consider a variant that appears in the desktop-based environment under two representations:

Representation	Pathogenicity Flag	Last Modified	Outcome
Representation A (%715C)	Likely Benign	13 June 2018	Not selected
Representation B (%D8BA)	Benign	9 September 2024	Selected in New Generation SOPHiA DDM™ ✓

Because Representation B was more recently modified (2024 vs. 2018), it is used as the source of truth in the New Generation SOPHiA DDM™. A system comment is appended to the annotation explaining this resolution, and the full history of both representations is visible in the Variant History section.

4. Ongoing process

To ensure that new conflicts introduced by SOPHiA DDM™ (accessible through the desktop), runs are handled consistently, we have implemented a nightly automated process that:

- Identifies any newly created variant conflicts arising from the desktop-based activity during the day.
- Applies the same most-recently-modified selection logic.
- Updates the corresponding annotation comment in New Generation SOPHiA DDM™ accordingly.

This means your web-based environment will always reflect the most current user evidence, without requiring any manual intervention on your part.

5. Our recommendations

To make the most of the improvements this update enables, we recommend the following:

1	**Annotate from New Generation SOPHiA DDM™ where possible** Performing user annotations in **New Generation SOPHiA DDM™** ensures a single, consistent record across all variant representations and contributes to more accurate variant statistics within your account and across the user community.
2	**Review your variant history if needed.** If you notice a change in a variant's annotation or an appended system comment, the **Variant History** section provides full context on what was consolidated and why.
3	**Request a detailed impact report if required.** If you would like a full list of the variants affected by this clean-up and the specific resolutions applied to your account, please raise a request via the Customer Portal and we will provide a tailored report.

6. Questions & Support

If you have any questions about this update or would like to discuss the impact on your specific workflows, please do not hesitate to contact us through the Customer Portal. Our team is available to assist you and provide any additional detail you may need.

Contact Us

Submit your query via the Customer Portal and a member of our team will respond promptly. For urgent matters, please mark your request as high priority.