

Automated reporting of negative results for the 100,000 Genomes Project

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Introduction

The South London GMC has recruited over 8,000 rare disease samples for the 100,000 Genomes Project (100KGP). A significant proportion of the probands (~40%) will not carry any tier 1 or 2 variants and will therefore be issued a negative result.

The GMC hub lab at Guy's Hospital are developing a solution for these negative cases using a fully automated system that will issue these results as soon as the patients come off the Genomics England pipeline, with no manual intervention required.

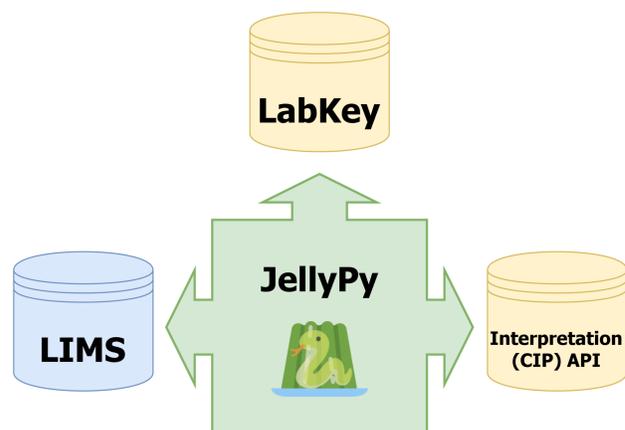
This solution has so far been implemented for Guy's and St Thomas' and effectively reduced the lab workload associated with these cases to zero.

Method

Genomics England (GeL) have developed and made available to GMCs the Interpretation API (aka CIPAPI), which holds variant and other information for 100KGP cases. Patient demographics are held in a separate system, LabKey. Both of these systems can be queried programmatically.

JellyPy¹ is a collaborative project between NHS bioinformaticians. It serves as a shared repository for code that interacts with the GeL infrastructure, and aims to reduce the duplication of work and produce consistency at a national level.

We developed an automated system for identifying and reporting negative cases programmatically (Figure 1). To achieve this we utilised functionality already available in JellyPy, and have contributed our own code back to the repository for other GMCs to use.



- 1 Find cases with no Tier 1/2 variants (JellyPy searches Interp-API)
- 2 Add cases & results to LIMS (import data from Interp-API)
- 3 Validate patient demographics (check LIMS against LabKey)
- 4 Close cases (create summary findings & submit exit questionnaire)
- 5 Create letter & email out (standard 100K template + summary findings)
- 6 Record activity, billing & close case in LIMS

Figure 1 – Automated negative reporting process at Guy's Hospital

For testing our workflow we made use of the CIPAPI beta environment provided by GeL. This allowed us to test our code without affecting live cases.

The code was developed following a thorough process of requirements gathering, architecture design, development, testing, code review and post-rollout monitoring. All code was developed in line with the ACGS best practice guidelines for bioinformatics.²

Outcomes

This solution has so far been implemented for Guy's and St Thomas' and effectively reduced the workload associated with these cases to zero.

This has allowed us to significantly increase the number of 100KGP cases being reported, with 300 results being issued this month. Clinical Scientist time is freed for interpreting other cases, and patients receive their results faster (Figure 2).

After implementation, we audited the first 150 cases and confirmed that this solution worked as expected.

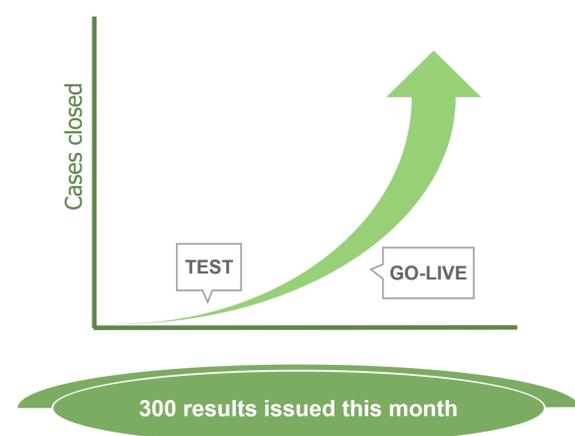


Figure 2 – Increase in cases reported since introduction of automated negative reporting.

Further Work

After a successful introduction in Guy's, we plan to roll out this system to other labs within the South London GMC.

We continue to contribute code to JellyPy to aid other GMCs wishing to implement similar solutions.

Since GeL will also be using the Interpretation API for the new Genomics Medicine Service (GMS), we hope that the system for automated reporting can be used beyond the 100KGP.

We are developing a system for importing variants and case details from GeL2MDT into our LIMS to allow interpretation of our in-house WES and WGS variants in one place.³

References

1. JellyPy - <https://github.com/NHS-NGS/JellyPy>
2. Guidelines for development and validation of software, with particular focus on bioinformatics pipelines for processing NGS data. <https://www.acgs.uk.com/quality/best-practice-guidelines/>
3. GeL2MDT - <https://github.com/NHS-NGS/GeL2MDT>