# **TA7:** Compare a decision support tool to a more traditional clinical decision-making approach, highlighting the benefits of the tool to clinicians and patients

**Introduction**

The analysis of data from NGS plays a crucial role in the workflow of the WMRGL. This document aims to compare a traditional approach to the analysis with the potential benefits offered by an integrated in-house DSS. The objective is to highlight the anticipated benefits of a DSS for both Clinical Scientists and patients, considering aspects like efficiency, accuracy and overall utility in a clinical setting.

**The Traditional Approach**

The Old-Style TSMP workbook, highlights a detailed but labour-intensive process for genomic data analysis. Genomic and Bioinformatic Clinical Scientists using this method would likely begin by manually collating various data outputs from bioinformatics pipelines. This involves gathering quality control metrics, extensive lists of genetic variants, detailed gene coverage information and other test-specific parameters, each often in separate files.

A significant portion of this traditional workflow would involve the manual review of these separate datasets. For instance, quality control would necessitate opening and interpreting individual files to determine the reliability of the sequencing run.

A core and particularly demanding component of the traditional method is the variant review. This necessitates carefully examining extensive datasets (spreadsheets) to complete the filtration of variants based on criteria such as read depth, allele frequency and quality scores. When performed manually, this filtration is not only time-intensive but also prone to human error and variability.

In order to determine each variant's clinical importance, the interpretation stage then necessitates manually cross-referencing it against numerous external databases and recent scientific publications. All these results are then collected into a clinical report.

**Streamlining Analysis with an In-House DSS**

An in-house Decision Support System would introduce features such as automation, integration, and intelligent analysis tools.

A DSS would offer a single, integrated platform in place of CS browsing separate data files. All relevant data, including variant calls, coverage information, quality control measures and related annotations, would be automatically processed and combined in one place. This removes the need to manually cross-reference many spreadsheets and enables faster evaluation of data quality. Variant filtering and prioritisation, which are frequently difficult tasks, would be much simplified as a DSS can be built with decision tree logic to automatically select variants. This allows CS to be able to concentrate their skills on a lot more manageable, highly relevant subset of variants, which would significantly decrease the manual strain. Furthermore, A DSS would also automate the annotation of variants using multiple datasets such as ClinVar. This gives users instant access, from within the system, to a multitude of data regarding the known associations, anticipated impact, and body of existing research for each variant, resulting in faster interpretation. Finally, the reporting process would become far more efficient as a DSS would be able to automate the generation of these clinical reports.

**Benefits for Healthcare Professionals and Patients**

The adoption of a DSS offers profound benefits. Forhealthcare professionals, who will be users of the DSS, the most immediate advantage is a significant reduction in the time and manual effort required for analysis and reporting. This efficiency allows them to dedicate more of their time to complex case review, direct patient care and research. The system's ability to consistently apply filtering rules, annotation sources and clinical guidelines also contributes to improved accuracy and reproducibility, minimising the risk of manual errors and variability. These improvements directly translate into benefits for patients. Faster analysis turnaround times mean quicker delivery of results, which can be critical for timely diagnosis and beginning appropriate treatment.

**Reflections on Implementing a Decision Support System**

The introduction of a DSS shifts the role of CS within the lab from being heavily involved in manual data processing to one of critical oversight and interpretation of clinical judgment. While the DSS is a powerful tool, it is designed to enhance, not replace, human expertise. Therefore, thorough validation of the system's algorithms, data sources, and outputs is essential. Clinicians require adequate training to understand the DSS's capabilities and limitations, ensuring they can use it effectively and critically evaluate its recommendations.