# **TA6:** Summarise a range of digital health tools intended for patient use providing an assessment of usefulness and provide a recommendation for applicability to a specific workflow

**Congenica**

Congenica is a clinical decision support platform designed for the analysis and interpretation of Rare Disease Genomic Data. Specifically, it is used to interpret rare disease sequence variants identified in Next Generation Sequencing analysis.

*Key Useful Functionalities:*

* Variant Classification: Utilises decision trees to automatically classify known variants, automation reduces analysis time, enabling faster turnaround for results.
* Phenotype-Driven Analysis: Integrates patient phenotype information to prioritise variants relevant to the clinical presentation.
* A screenshot of a computer

  AI-generated content may be incorrect.Comprehensive Reporting: Generates reports suitable for clinical decision-making.

Figure 1: Congenica Rare Disease Dashboard Interface.

This is the main patient dashboard within the Congenica platform, used for genomic analysis in rare disease diagnostics. Each row represents an individual patient case, summarising key metadata including analysis status, gene panel used, CNV analysis completion, protocol, sample type and report availability. Users can filter, search and access detailed clinical reports directly via the "View report" button. The dashboard facilitates streamlined tracking and management of large-scale rare disease genomic workflows.

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Figure 2: Patient Report View in Congenica Rare Disease Platform.

This figure displays the patient-specific report interface generated by the Congenica platform. It summarises critical clinical and genomic metadata, including lab ID, reported and genomic sex, phenotype summary, gene panels applied, CNV analysis status and sequencing protocol. This structured report enables clinical scientists to interpret findings within the full clinical context, supporting accurate and timely diagnosis.

**Applicability to Childhood Solid Tumour Panel workflow:**

This workflow involves identifying and interpreting genetic variants in pediatric patients with suspected solid tumors, using panels like CSTP (Childhood Solid Tumor Panel). Variants are identified through Sanger, NGS or MLPA and processed through Congenica.

Congenica plays an important role in the workflow by allowing variant evidence for CSTP-only genes to be recorded directly within the system. This means there’s no need to create or manage separate external forms. For genes that are also relevant to other clinical areas, like BRCA, APC, or CDH1, Congenica makes it easy to link in variant forms created outside the platform, so everything stays connected. It also supports tailored reporting templates for different diseases, which helps ensure reports are consistent, accurate, and clinically useful.

**Reflection - Congenica**

After reviewing Congenica, I appreciated how purpose-built it is for rare disease diagnostics. Its deep integration of phenotype-driven analysis and the ability to directly capture variant evidence made it clear how much time and effort it can save in a clinical workflow. Seeing how it supports disease-specific templates and allows streamlined report generation showed me just how powerful structured genomic interpretation tools can be when embedded into the diagnostic process. However, I also noticed that the user interface, while informative, might require a learning curve, particularly in understanding how to navigate patient metadata and link external variant forms.

**Alissa Interpret**

Alissa Interpret is a platform developed for the interpretation and reporting of genomic variants derived from NGS data. It is designed to aid in the analysis phase of genomic workflows, facilitating efficient variant analysis.

*Key Features:*

* Variant Analysis: Supports the interpretation of various genomic alterations, including single nucleotide variants (SNVs), insertions/deletions (indels), copy number variations (CNVs), loss of heterozygosity (LOH) and gene fusions.
* Automated: Provides version-controlled pipelines that automate variant filtration, annotation, classification and report generation.
* Phenotype Prioritisation: Integrates patient phenotypic information to prioritise variants based on relevance.
* Databases: Access to both internal and external variant databases to inform variant interpretation.
* Generates Reports: Generates reports that can be tailored to specific laboratory requirements, ensuring clarity and compliance with standards.

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Figure 3: Alissa Interpret Main Dashboard.

This image shows the file management interface within the Alissa Interpret platform. It provides a view of uploaded variant call format (VCF) files, including key metadata such as genome build (GRCh38), lab result status and total variant count. Clinical Scientists can filter, search and manage files efficiently.

**Applicability to TSO500 and Pan-Haem workflow:**

In the TSO500 workflow, Alissa is used not only for variant assessment, but also forcoverage checks, peer-reviewed commenting and final report generation.  
Each variant is reviewed, annotated and discussed by two checkers directly within the platform. Annotations often include links to reference databases, artefact exclusions and notes on known polymorphisms. Following this, a structured PDF report is generated using a template.

We also rely on Alissa’s ability to manage custom gene panels, classification tree logic and manual overrides, all of which are reflected in the final report. If any of these processes are done outside of an integrated platform, we risk increasing turnaround time and compromising data integrity.

Alissa also plays a central role in the Pan-Haem pipeline. The analysis starts with sequencing data processed via Illumina’s DRAGEN pipeline. The resulting VCF files are uploaded into Alissa. Alissa then uses classification trees tailored for SNVs, CNVs, and structural variants based on the national Cancer Test Directory. These trees help prioritise variants according to clinical significance, which is critical for accurate diagnosis and treatment guidance.

**Reflection - Alissa**

Alissa Interpret offered a more flexible and lab-facing interface, especially well-suited for labs managing a high volume of files across various genomic indications. I was impressed by the detailed file tracking and variant management system, and how well it integrates with broader workflows like CNV analysis. It appears more customisable, which benefit labs like WMRGL that handle both germline and somatic variant interpretation. That said, it seemed slightly more technical and possibly less streamlined for clinical scientists who need fast insight from an output.

**Final Reflection**

After reviewing both Congenica and Alissa Interpret, I realised there are several key areas where I need to develop my knowledge and skills further to use these tools effectively in clinical or diagnostic settings. While I could follow the interface and general functionality of both platforms, I found that I lack a deeper understanding of variant classification frameworks, particularly how the ACMG guidelines are practically applied within these tools.

I also recognised a gap in understanding how to evaluate when to use one tool over another based on diagnostic needs, lab priorities or reporting requirements. This highlighted the importance of clinical judgement and systems thinking when integrating digital tools into practice.

**Action Plan for Knowledge gaps**

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| Goal | Action | Resources |
| Build competence in variant interpretation using ACMG guidelines | Complete a short course or internal training module focused on ACMG classification and variant curation | Genomics Education Programme, ESHG courses, internal SOPs |
| Understand strategic tool selection for different workflows | Review real-world case studies and participate in workflow planning meetings where tool use is discussed | SOPs, attending lab/clinical discussions |
| Improve ability to communicate genomic results | Practice summarising variant interpretations into clear, clinical-facing reports with supervision | Support from senior clinical scientists or bioinformaticians |
| Learn how digital tools are integrated into wider clinical systems | Attend IT and digital transformation briefings (where available), and explore LIMS/EHR integration pathways | Hospital IT teams, digital strategy groups, documentation reviews |
| Consolidate learning through documentation and reflection | Create a short comparison guide or visual workflow showing how Congenica and Alissa fit into specific clinical contexts | Personal notes, supervisor feedback, platform user guides |