Digital Health Tools & Clinical Decision Making

**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

## Activity

### Part 1 – background information

Digital heath tools:

* Alissa – Cancer services
* Congenica – rare diseases
* You should have access to both systems
* Find and refer to the relevant SOPs on iPassport:

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### Part 2 – Activity:

#### “Evidence the activity has been undertaken by the trainee​.”

Summarise the Alissa and Congenica digital health tools.

* Describe each tool and state what they are used for.
* How useful are they?
* Recommend applicability to a specific workflow with reasons (i.e. why).
* Can include screenshots.

#### “Reflection on the activity at one or more time points after the event including learning from the activity and/or areas of the trainees practice for development.”

* Reflect after reviewing the 1st tool
* Reflect after reviewing the 2nd tool
* Reflect at end of TA

#### “An action plan to implement learning and/or to address skills or knowledge gaps identified.”

* Include this at the end.
* Describe each tool and state what they are used for.
* How useful are they?
* Recommend applicability to a specific workflow with reasons (i.e. why).
* Can include screenshots.

**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.
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  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's Role in the Workflow:*

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.

*Reasons for Applicability:*

* Centralises and standardises variant interpretation, improving turnaround time and reducing human error.
* Supports complex, multi-source workflows by allowing integration of both internal (Congenica) and external (variant forms) data.
* Customisable per disease area, with specific templates aligning to clinical guidelines.

**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.

**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.