INTRODUCTION

Everyday discoveries in pediatric genomics create opportunities to understand and treat rare genetic diseases. Precision management of rare diseases demands business process automation beyond the four walls of the lab. Transforming pediatric healthcare through genomic sequencing requires more than discovery; genomic technology and bioinformatics make these processes fast, easy, and routine.

METHOD

L7 Informatics worked with Rady Children’s Institute for Genomic Medicine (RCIGM) to implement the Enterprise Scientific Platform (L7|ESP) to break down data silos, virtualize, and digitalize RCIGM precision medicine workflows across departmental and organization walls leading to better outcomes.
RESULTS

L7|ESP, a sample-centric platform, breaks down data silos between clinical orders, EMR, wet lab, sequencing lab, bioinformatics compute lab, and cloud-based interpretation services (AI/ML). This created end-to-end process automation that increased quality, workflow efficiency, and throughput with decreased turnaround times.

Additionally, L7|ESP created a collaborative data management platform across physicians, lab scientists, clinical operations, genomic experts, and clinical research at RCIGM, increasing workflow efficiency, data integrity and catalyzing scientific insight while decreasing regulatory risk and TAT.

L7|ESP enabled process changes and quality improvements such as the WGS Assay assembled from re-useable protocols (Integrated Instrument Protocol, LabProtocol,Analytical Protocols; see Fig 1).

![Fig 1. Clinical sequencing workflows. Integrations include instruments from Illumina, Thermo Fisher, and Roche. Computational workflows upload results to L7|ESP.](image)

CONCLUSION

By digitalizing, automating, and simplifying all laboratory processes on a single regulatory compliant informatics platform with apps and instrument integrations, a unified platform for managing operational data and processes is created.

L7|ESP is such a platform that increased workflow efficiencies and throughput to support RCIGM’s efforts to expand access to genomically informed Rapid Precision Medicine nationwide for children suffering from diseases of unknown origin.