DELIVERING NEWBORN SCREENING RESULTS TO PHYSICIANS USING FHIR

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Abstract

Newborn screening (NBS) is a public health initiative with the goal of identifying infants with life-threatening but treatable disorders before the onset of symptoms. Each year in the US, NBS detects thousands of affected infants. The current NBS panel includes a number of disorders (e.g. cystic fibrosis, PKU, galactosemia) that are primarily screened for using biochemical assays however newer disorders included on the panel lack robust biochemical markers (e.g. pompe disease, MPS I) for detection and may rely on genetic testing for confirmation of the disorder. In cases in which an infant screens abnormal for a disorder, they are referred to a clinical specialist (e.g. pediatric pulmonologist) for confirmatory testing, diagnosis, and treatment. Recently, the Utah NBS Program developed and is piloting a universal exome-based method for secondary screening. Ideally, the details of the results would be shared with the clinical specialist along with the biochemical NBS results to help them with clinical decision making. However, there is currently no mechanism for sharing and utilizing these results beyond a PDF-based format which is how NBS results are reported to clinicians. The goal of this project was to develop a prototype application for the reporting and reuse of NBS genomic variant results to clinical specialists using Fast Healthcare Interoperability Resources (FHIR). FHIR is a healthcare standard that represents specific data artifacts found in typical clinical scenarios (Patient, Condition, Encounter, etc.) as standardized "resources". Resources are linked together via identifiers to provide a standardized and robust representation of clinical events. This integration of genetic information from newborn screening into point-of-care medical treatment is part of an initiative known as Sync for Genes. This project developed a pipeline using Python to transform NBS results into FHIR resources that can be continuously analyzed by clinical specialists. The pipeline was then tested on the NBS results of a hypothetical cystic fibrosis positive infant and proven to be effective in generating an application oriented towards clinical specialists that displays FHIR data. The physician-facing prototype is based on another prototype application, ResultsMyWay, which is geared towards the delivery of NBS results and information to parents. Next steps for this project include the incorporation of this method into ResultsMyWay as the provider-facing portal. This type of application that allows for electronic reporting and reuse of data has the potential to improve the care of newborns.
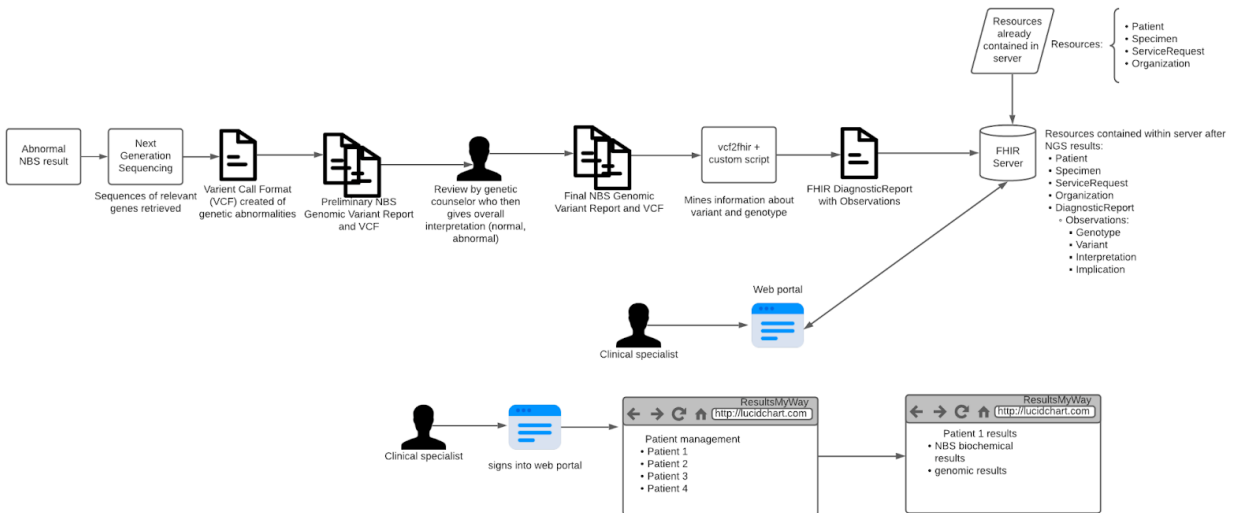
Figure 1: Newborn Screening with FHIR Pipeline

Figure 2: Original Form of Results Report Compared to FHIR Resource

DISORDER/*TEST*	DATE TESTED	RESULTS	DETERMINATION/ *NORMAL RANGE*
Cystic Fibrosis *CFTR DNA*	10/16/2018	Abnormal	**ABNORMAL**
Congenital Hypothyroidism *TSH*	10/15/2018	7.7 µIU/mL	Normal < 20 µIU/mL
Biotinidase Deficiency *Enzyme activity*	10/15/2018	Normal	
Congenital Adrenal Hyperplasia * *17-OHP ELISA*	10/15/2018	5.4 ng/mL	
Cystic Fibrosis *Immuno-reactive Trypsinogen ELISA*	10/17/2018	344.9 ng/mL	
Acylcarnitine Disorders *MS/MS Tandem Mass screening*	10/15/2018	Normal	
Amino Acid Disorders *MS/MS Tandem Mass screening*	10/15/2018	Normal	

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    },
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    }
  ],
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