OptraHealth
Engineering Digital Healthcare

IPHRONESIS FOR NEXT GEN CLINCIAL GENOMICS

ABSTRACT

OptraHEALTH™ presents **iPhronesis™: Machine learning based BigData Analytics Platform** with knowledge automation engine **for Precision Medicine.**

iPhronesis™ is built specifically to address the growing needs of the biomedical community around working with BigData that includes access to bioinformatics/ clinical/ experimental databases, integration with disparate data sources, interactive workflow designer, powerful analytics engine deployed on cloud architecture and using BigData environment and visualization tools for quick, accurate and innovative ways to visualize data. We aim to empower our users to generate interpretive insights into complex datasets simply and easily.

IPHRONESIS™ MODULES

The following is a list of iPhronesis™ modules that are readily available.

DATABASES

iPhronesis™ provides many different databases for several industry segments. Each database undergoes a manual schema curation process wherein we add additional information to help users understand the data better. EMR data is acquired by EMR connectors that we have developed that work with HL7 and CCDA data streams. For unstructured data such as from experimental databases we use machine learning techniques to map schemas and entities. Each mapped entity undergoes manual validation process. Following is a list of few databases which are currently available.

Databases			
Clinical	**Genomic**	**Proteomic**	**Others**
ADNI	1000 Genomes	BIND	PubMed Central
Ovarian Cancer	ClinVar	MINT	SIDER
AHA Cardiovascular Genome	dbGAP	PRIDE	GEO
Phenome Study (dbGAP)	dbMHC	GelMap	ClinicalTrials.gov
MIMIC2	dbSNP		
EMR Systems	PGP		
	COSMIC		
	ArrayExpress		

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The following is a list of EMR connectors that are currently available.

EMR Connectors			
EPIC(open.epic.com)	AllScripts(AllScripts Dev)	VistA(Open Source)	Practo

WORKFLOWS

iPhronesis™ provides a workflow module that allows users to quickly and efficiently define workflows which in turn use algorithm to address specific functionality. The workflow editor is implemented in HTML as canvas object and allows for smooth drag drop etc. Nodes represent algorithm, users choose from a wide variety of algorithms depending on the use case.

The following is a list of algorithms available for genomics applications.

Core Genome Analysis		
Aligners	Variant Detectors	Annotators
BWA (bwa-mem, bwa- aln) BowTie2 Stampy STAR Segemehl SOAP	GATK SamTools SOAP2 MaCH CNVnator cn.MOPS VarScan	SNPEff ENSEMBL VEP UCSC Variant analysis tool FunSeq
Pipelines		
Ruffus Luigi	Cloud Scaler	HDFS
Machine Learning Algorithms		
		Tools implementing ML
		GATK SomaticSeq VAAST

VISUALIZATIONS

iPhronesis™ provides a suite of visualization widgets that make view complex datasets simple. The selection of a visualization widget is determined when the workflow is designed but may be optionally changed when viewing the data. iPhronesis™ intelligently detects is a certain data view is allowed for a certain widget and only shows compatible widgets for user to view.

The following visualization widgets are currently available.

Visualization		
Core Genome	*Others*	
Chromosome plot, segment and normalization plots Variation viewer Data tables with sort, order and tag functionality Concept diagrams HeatMaps Cluster Maps Network diagrams		Bar charts Line charts

All visualization widget uses the latest charting libraries and several are custom built. This approach allows for viewing data in a very precise manner and client side rendering takes the user experience to the next level.

INTERPRETATIONS

A workflow is only complete when it delivers powerful interpretations. iPhronesis™ provides industry standard workflows for various industry segments to get you quickly up to speed and instantly start using validated, approved and performance tuned workflows leading to high interpretative power. In clinical genomics applications such as workflow is implemented to ascertain pathogenicity of detected variants and map population statistics values such as odds ratios, p- values and relative risks to generate lifetime risk assessment. For the simpler inheritable disorders, iPhronesis in addition to the former also provides literature evidences that helps in accurate diagnosis and in genetic counselling.

Following is a list of ready-to-go domain workflows available in iPhronesis™.

Genome Workflows	***Others***
GATK Best Practices Cancer Mutation Detection CNV Analysis Multifactorial Risk Assessment for select phenotypes.	

KNOWLEDGE AUTOMATION

The process of interpretations is only complete when concrete scientific evidences are provided. At times such data is buried un structured data and is not semantically organized. iPhronesis™ bridges this gap by providing a very powerful module that ingests data from scientific literature and contextually connects literature with interpretations such as drug- gene interactions, drug signatures or structural activity of drugs. This allows for rapidly identifying targets of drugs of interest, identifying false positives, ascertaining efficacy and potency in comorbid conditions etc. Such evidence based insights are available in iPhronesis™, additionally, there is also a BioNLP (Bio Natural Language Processing) interface that allows users to compose queries in natural language, parsing which iPhronesis™ provides results.

For more information and demo please contact us today at info@optrahealth.com

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