MED AVANT

AND

PhenoTips

INTEGRATING GENOTYPE AND PHENOTYPE DATA

Michael Brudno

Department of Computer Science & Donnelly Centre

University of Toronto

Centre for Computational Medicine

Genetics and Genome Biology

Hospital For Sick Children

FORGE CANADA CONSORTIUM

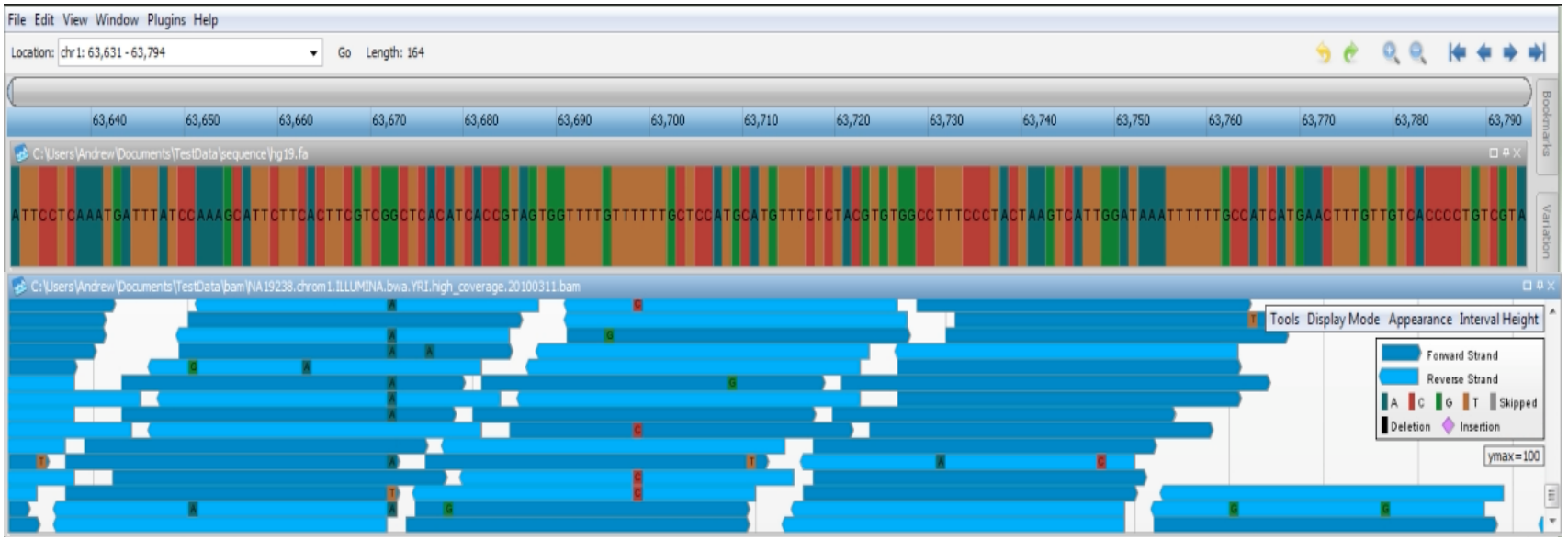
From Genome to Genomic Medicine

- shift: **interpretation and analysis** challenge
- genome browsers made for single genomes, not cohorts
- disconnect between tools used for data analysis and visualization

Marc Fiume

SAVANT & MEDSAVANT: BROWSERS FOR MEDICAL SEQUENCING

Savant Genome Browser



- Dynamic Visualization of HTS Datasets (BAMs, VCFs, etc.)
  - Local or Remote
- Integration with External Databases (e.g. UCSC)
- Plugin Framework Allows for Extension

Savant Plugin Framework

- **unlocks the potential for performing visual analytics**
- **mutually beneficial** for both users and tool developers

  for users: perform complex data analyses on-the-fly within a visual environment

  for programmers: platform for simple development and deployment of new tools

- Some Savant Plugins

  - RNA-seq analyzer, EdgeR Differential Abundance Estimator, Application Wrapper, GO Navigator, WikiPathways Navigator, Data Table… (16 and counting)

MedSavant: Where to look?

SAVANT

MEDSAVANT

Database

Contents

- patient: e.g. age, gender, family
- phenotype: e.g. symptoms and scores
- genotype: e.g. variants

Query Engine

Filters

- set of user specified rules that **remove low quality or uninteresting variants** on the basis of:
  - basic patient data, phenotype data, genotype

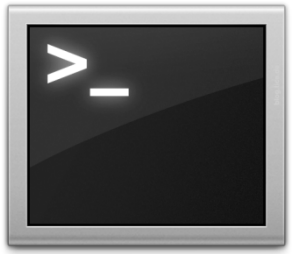
Visualization Framework

Charts and Tables

- graphical presentation of trends in the data:
  - **summary** : histograms, pie charts
  - **aggregate** : over genomics regions, ontology terms

Tools for processing genetic variants

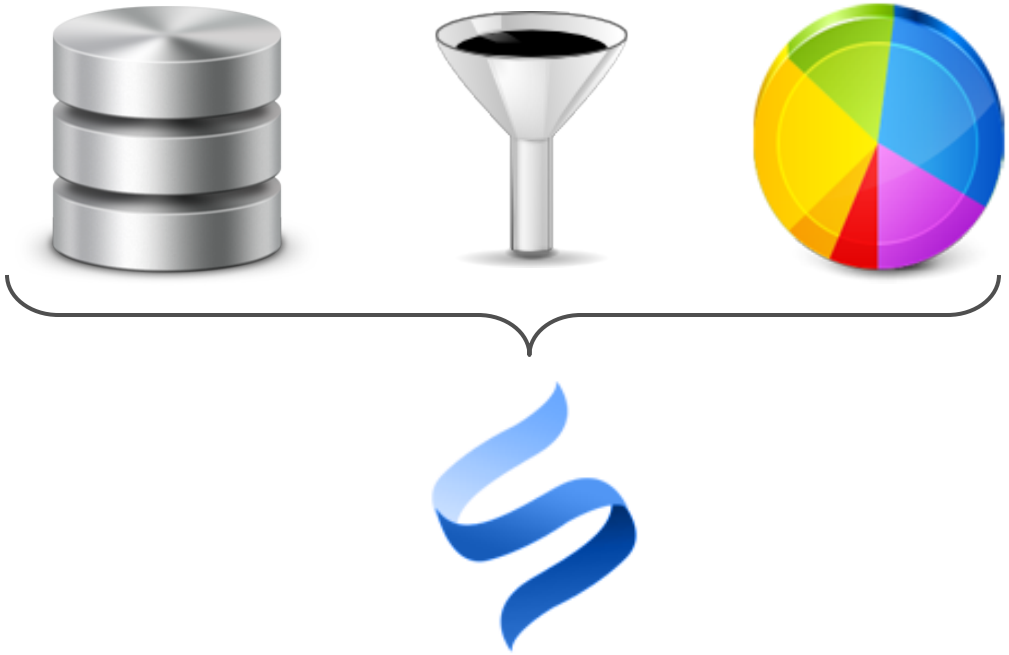




- powerful
- expert use
- difficult to maintain

- not as powerful
- easy to use
- difficult to maintain

Introducing MedSavant



- powerful
- easy to use
- easy to maintain

DEMONSTRATION

Conclusions

- MedSavant is a platform for **efficient storage, filtering, and visualization** of genetic variants

- **Accelerating the discovery of causal variants** in disease sequencing studies

- Plugin framework enables **integration of cutting-edge tools for genetic variant analysis**

http://medsavant.com/

Marta Girdea

PHENOTIPS: PATIENT PHENOTYPING TOOL AND DATABASE

Phenotyping for FORGE

- Rare Disorders Are not Always Named
  - Important to collect phenotype data to cross-reference against cases observed across the country
  - Currently Two Alternatives: free text or checkboxes

Dysmorphic features
- df
- dysmorphic
- dysmorphic faces
- dysmorphic features

Congenital malformation/anomaly:
- congenital anomaly
- congenital malformation
- congenital anamoly
- congenital anomly
- congential anomaly
- congentital anomaly
- cong. m.
- cong. Mal
- cong. malfor
- congenital malform
- congenital m.
- multiple congenital anomalies
- multiple congenital abormalities
- multiple congenital abnormalities

Phenotypic description (Clinical symptoms)

Behavior, Cognition and Development
□ Global development delay
□ Fine motor delay □ Gross motor delay
□ Language delay
□ Learning disability
□ Mental retardation
□ Mild
□ Moderate
□ Severe
□ Attention deficit hyperactivity disorder
□ Autism
□ Pervasive developmental delay
□ Psychiatric disorders (Specify below)
□ Other: _____

Neurological
□ Hypotonia
□ Seizures
□ Ataxia
□ Dystonia
□ Chorea

Cardiac
□ ASD
□ VSD
□ AV canal defect
□ Coarctation of aorta
□ Tetralogy of fallot
□ Other: _____

Craniofacial
□ Craniosynostosis
□ Cleft lip □ Cleft palate
□ Microretrognathia □ Retrognathia
□ Facial dysmorphism (Specify below)
□ Other: _____

Eye Defects
□ Blindness
□ Coloboma
□ Epicanthus
□ Eyelid abnormality (Specify bellow)
□ Other: _____

Examples of lists:
* dd. cong. malfor. behav. pro.
* dd. mental retardation
* df< delayed puberty
* df& lt
* dd df mr
* mental retar.short stature

What about Ontologies?

- Extensive Ontologies Describe Human Phenotypes:
  - London Dysmorphology Database
  - Human Phenotype Ontology
  - SNOMED
- Initial Reaction from Clinical Geneticists:
  - Absolutely not, too complicated
- So Can We Make it Simple?

DEMONSTRATION

PhenoTips

- All FORGE patient phenotypes will now be recorded through the web UI (NO PAPER!)

- Working to install similar database at SickKids for Clinical Geneticists and Molecular Diagnostics

- Simple User Interfaces can make (even) Clinicians use Computers ☺

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http://genomesavant.com/med

PhenoTips Team

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http://phenotips.cs.toronto.edu