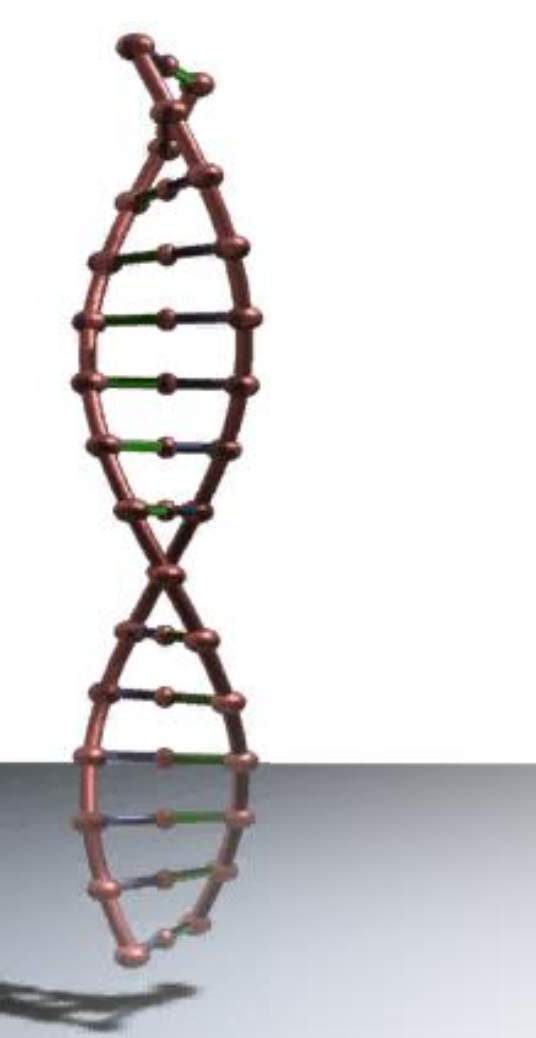

Shifting the Paradigm for Genetic Sample Resources

J. Claiborne Stephens, CEO, Genomics GPS
Ron Bromley, Ena Bromley, BSSI
Ted Kalbfleisch, Intrepid BioInformatics

Disclaimer

- Genomics GPS is focused on the strategic application of genomic studies, samples, and analyses in a variety of commercial applications
- Clay Stephens, Founder and CEO, is currently the only employee

Overview

- Current Process
- Proposal for Change
- Contrast
- Scientific Benefits of New Model
- Commercial Benefits
- Discussion

Genetic Biosamples

- Typically blood
  - Treated for clotting
  - Antimicrobial
  - Frozen
- Extracted DNA
  - Frozen
  - Blotted
- Desired data
  - Genotype or sequence of one or more loci

Current Model

Sample is

- ✓ Taken and prepped for storage
- ✓ Retrieved and prepped for genotyping
- ✓ Genotyped
- ✓ Remainder returned to storage

Data obtained

Inventory (where is it, how much is left, what format is it in)

Proposed Model

Sample

- ✓ Taken and prepped for genotyping
- ✓ Uniformly genotyped and imputed
  - Note: combination of direct and indirect methods
- ✓ Remainder sent to storage (archival)

Data obtained

Inventory (where is it, how much is left, what format is it in)

Pros and Cons of Current Model

Positives

- Familiar
- No upfront genotyping expense
- Obtain only the data desired
- Genotyping/sequencing costs continue to drop

Negatives

- Possibility that sample gets lost or consent expires
- **Expense** in storing/retrieving/ prepping sample
- **Time lag** in above
- No/Limited use of sample
- Non-uniform data generated

Pros and Cons of New Model

Positives

- Data available and uniform
- Reduced expense for storing/ retrieving/prepping sample
- Reduced time lag in above
- Maximize use of sample
- Sample loss avoided
- Data may improve over time as reference panels and imputation algorithms improve

Negatives

- Upfront genotyping expense
- May still miss the desired data (e.g., sequence)
- Infrastructure for data maintenance and access may be lacking

Perfect Genomic Data

- Complete and accurate full sequence
- Including simple structural (indel) variation
- Including complex structural variation (CNV)
- Including phase

- Exclusions
  - Mosaicism
  - Epigenetic phenomena

What is Sacrificed in Proposal?

- *de novo* mutations (single patient)
- Private variation (single family)
- Rare variants that fail to impute
- Common variants that fail to impute
- Indels and CNVs that fail to impute

- Phase should be accessible as part of imputation process

Commercial Considerations

Per Sample

- Incurring upfront genotyping costs in place of sample prep/storage/retrieval costs
- Storage and retrieval is now data, not sample

Infrastructure

- Freezers and personnel now minimized to storage of archival (non-frozen?) samples
- Shift to data infrastructure

Discussion

Thanks!

The End(s)