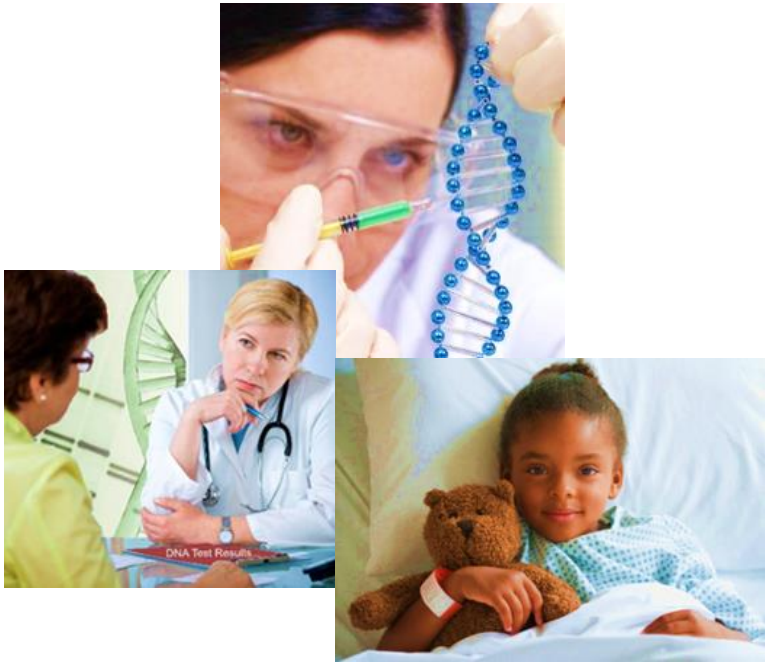
Introducing VarSeq:

Variant Discovery & Gene Panels Made Easy

Gabe Rudy with intro from Andreas Scherer

October 1, 2014

We are building the MRI for the genome!

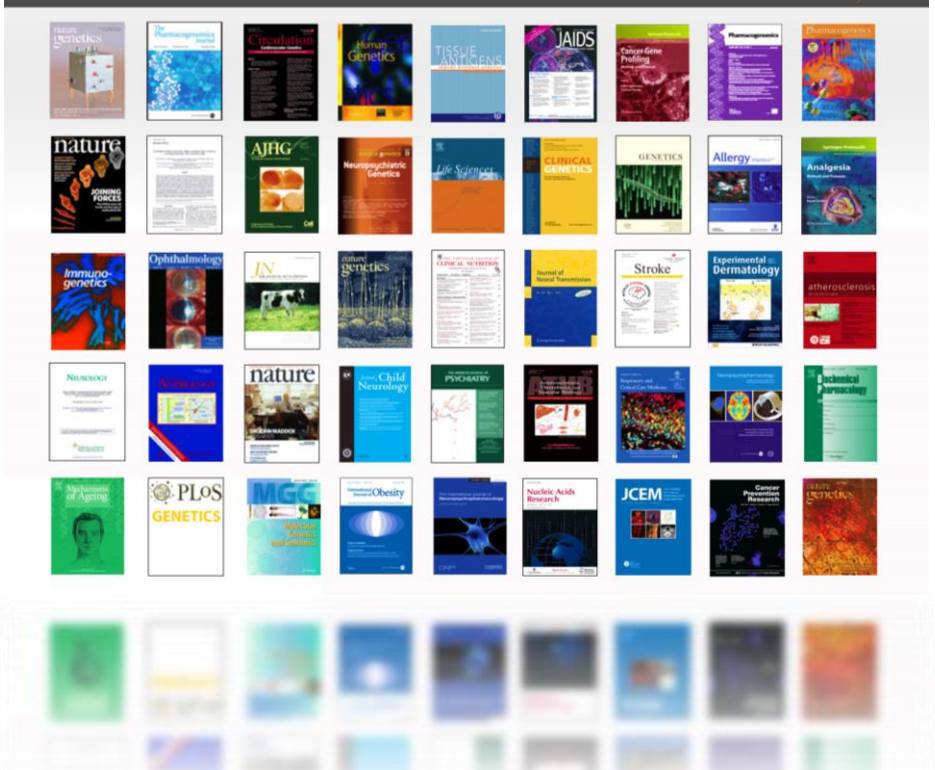


The analysis of genomic data and the prediction of outcomes reshapes the way we diagnose disorders, predict outcomes and select the best possible care options for a patient.

Thousands of Users in Research and Translational Labs

Hundreds of Customers Across the Globe

Over 850 Citations



Genetics Adoption Curve

Early Stage	Moderate Adoption	High Adoption
Market focus is on science and research, lack of infrastructure, clinical evidence and physician education.	Clinical genetic standard for selected targets and therapeutic areas. Bioinformatics increasingly crucial for diagnosis and treatment selection.	Greater availability of data around testing with genetic services becoming standard of care for a majority of patients.

Regulatory Landscape	Reimbursement	Bioinformatics
Testing Technology	Physician Adoption	Consumer Demand

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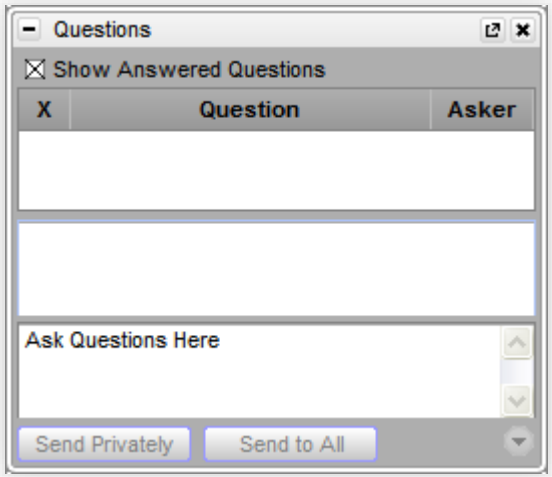
Gabe Rudy
VP of Product Development
October 1, 2014

Agenda

1. Roots of VarSeq
2. Demo
3. What's Next at Golden Helix
4. Questions

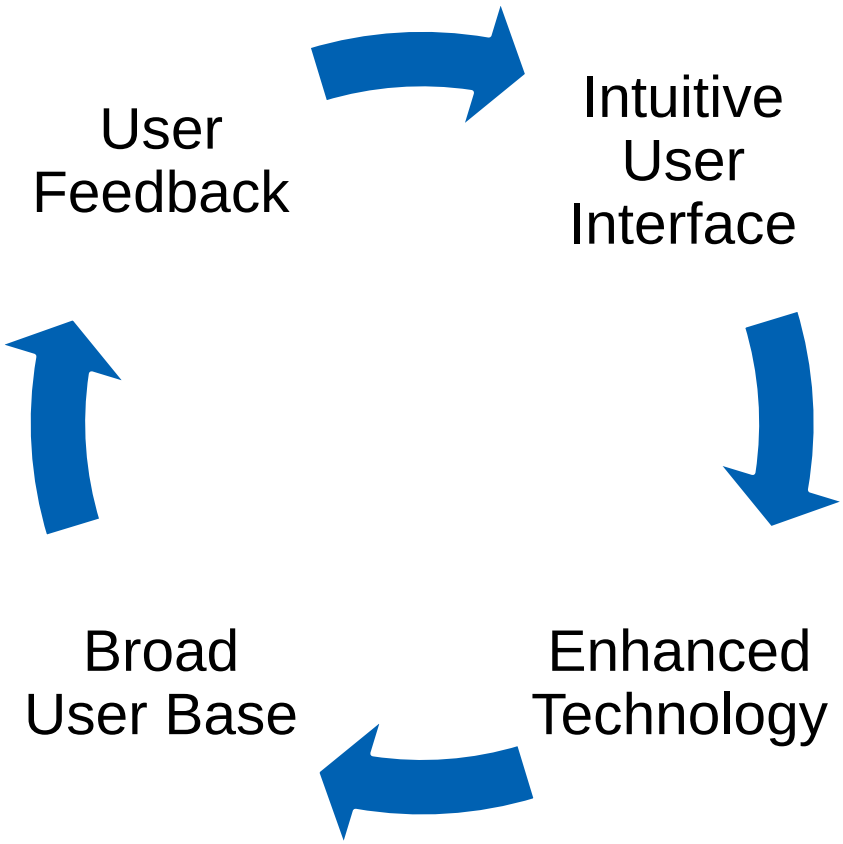
Questions During the Presentation

Use the Questions pane in your GoToWebinar window

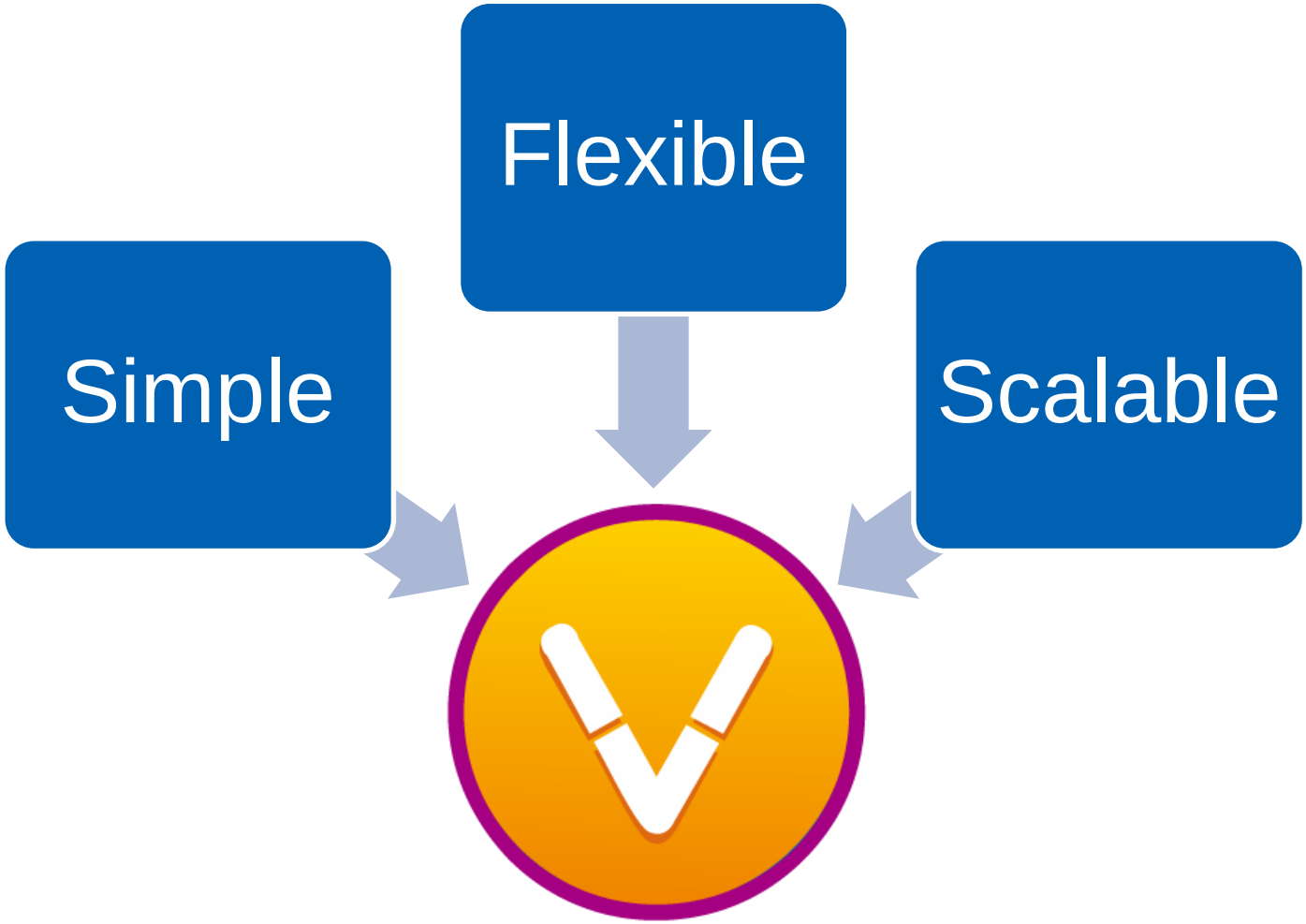


Built on Strength

- Experience empowering users to solve hard problems
- Tight feedback loop with users and stakeholders
- Support users with licensing models aligned with customer interests
- Evolve and share core technologies across products



Showing not Telling

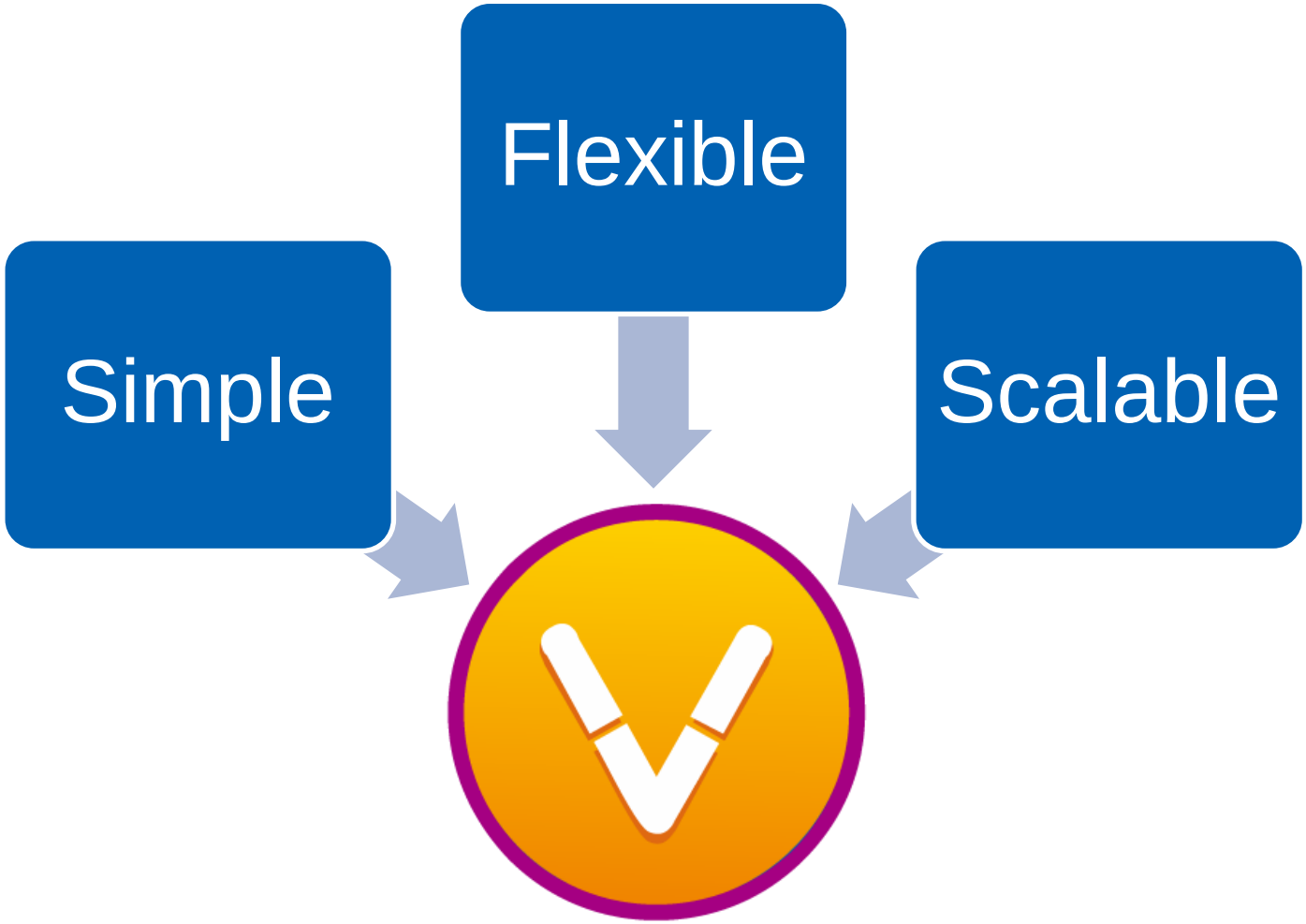


Simple

- Get started easily
- Utilize pre-built best practice workflows
- Analyze samples in batches
- Focus workspaces to accelerate sample turn-around

[Demonstration]

Showing not Telling

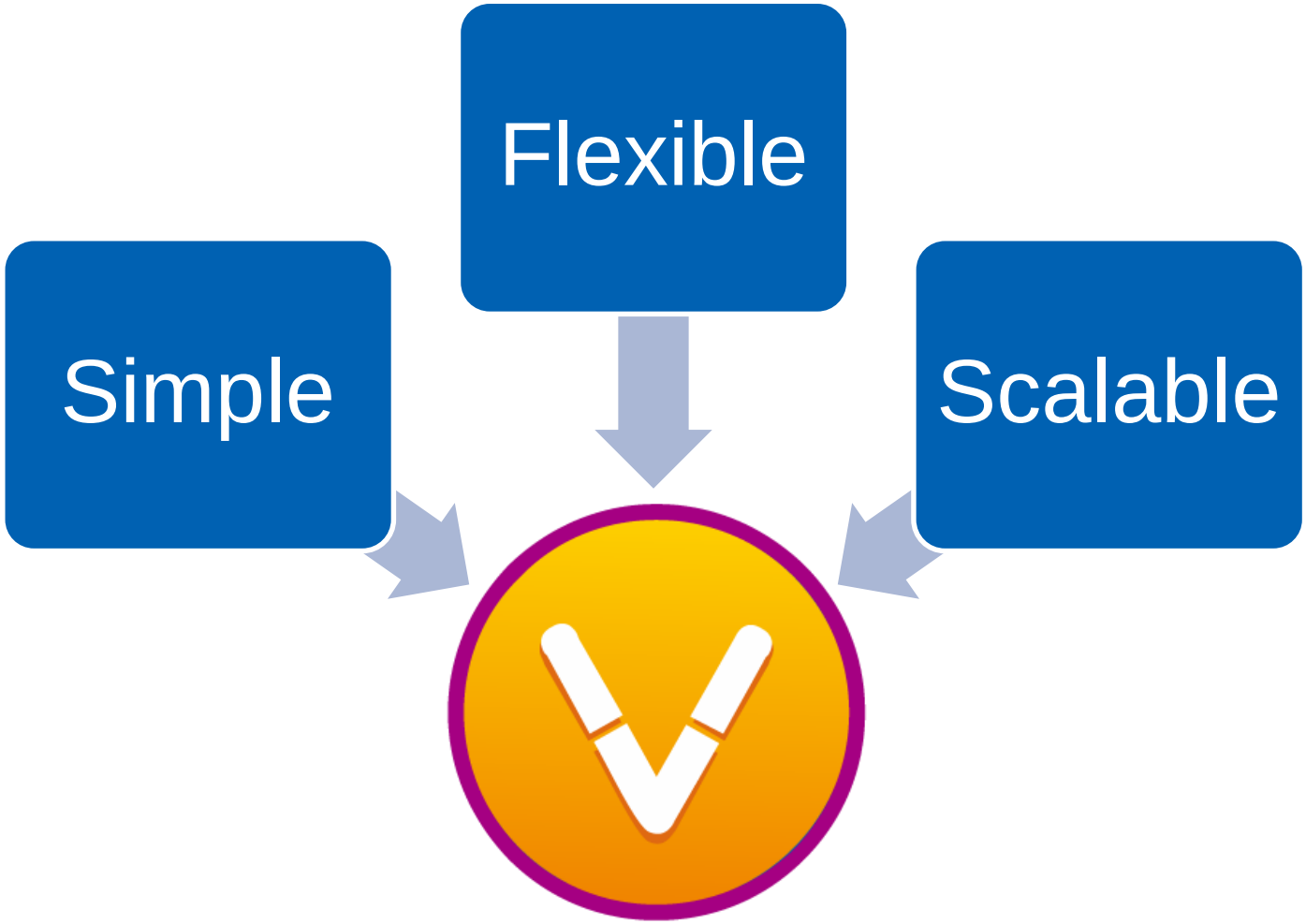


Flexible

- Drilldown with context
- Annotate from extensive library of public databases
- Filter on any field of a data source
- Visualize sample and public data with GenomeBrowse
- Arrange quadrants to support multiple vantage points
- Document insights with supporting evidence

[Demonstration]

Showing not Telling



Scalable

- Analyze millions of variants
- Investigate multiple hypotheses in tandem
- Validate robustness of filtering thresholds
- Export results to VCF, text, and multi-tab Excel files

[Demonstration]

Acknowledgements

- Stakeholders
  - Jason Byars
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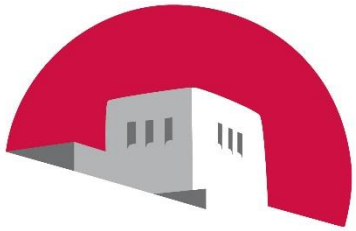
  - Ken Kaufman
  - Sam Strom
  - Jeff Moore
  - Jeff Rosenfeld
  - Scott Ness



Liverpool Women's NHS Foundation Trust

UNM

FLUXION

UCLA Health

Royal Surrey County Hospital NHS
NHS Foundation Trust

Cincinnati Children's®

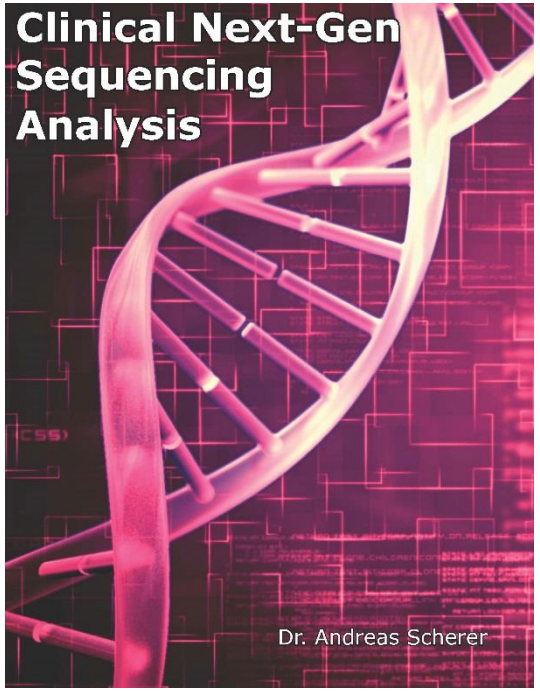
ILLINOIS
UNIVERSITY OF ILLINOIS AT URBANA-CHAMPAIGN

October

varSEQ™

Announcement

ASHG 2014
San Diego, CA | October 18-22
ashg.org/2014meeting

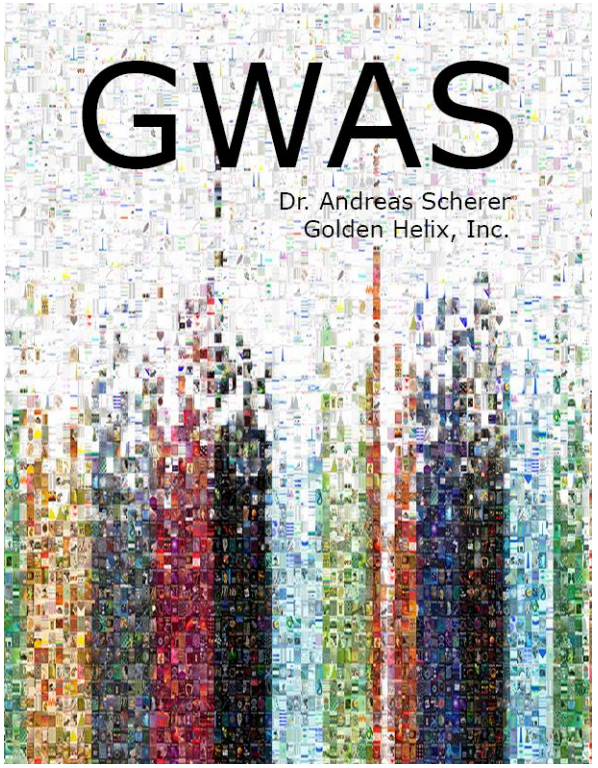


End of 2014

varSEQ™

Commercially Available

Golden Helix gives back



Q1 of 2015

varSEQ™

More features and capabilities

SNP & VARIATION SUITE

Release of 8.3:
More Genomic Prediction,
More Methods to Study Complex Diseases

Molecular Med TRI-CON

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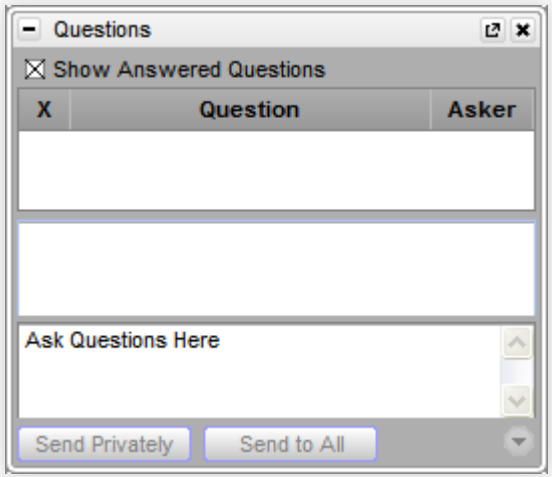
Do You Have Any Questions?

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