Tips and Tricks for Genomic Analysis in SVS

August 6, 2014

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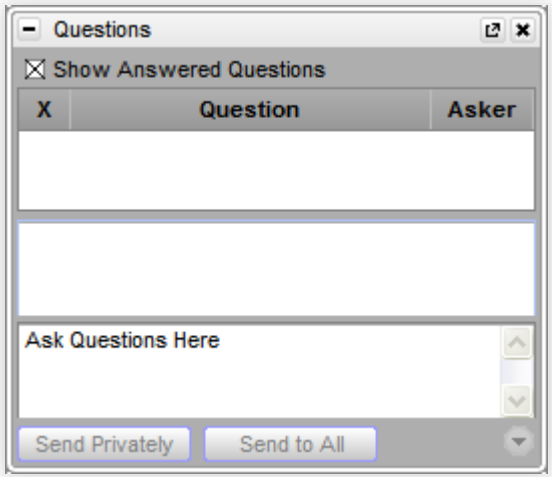
Field Application Scientist

GOLDEN HELIX
Accelerating the Quest for Significance™

Questions during the presentation

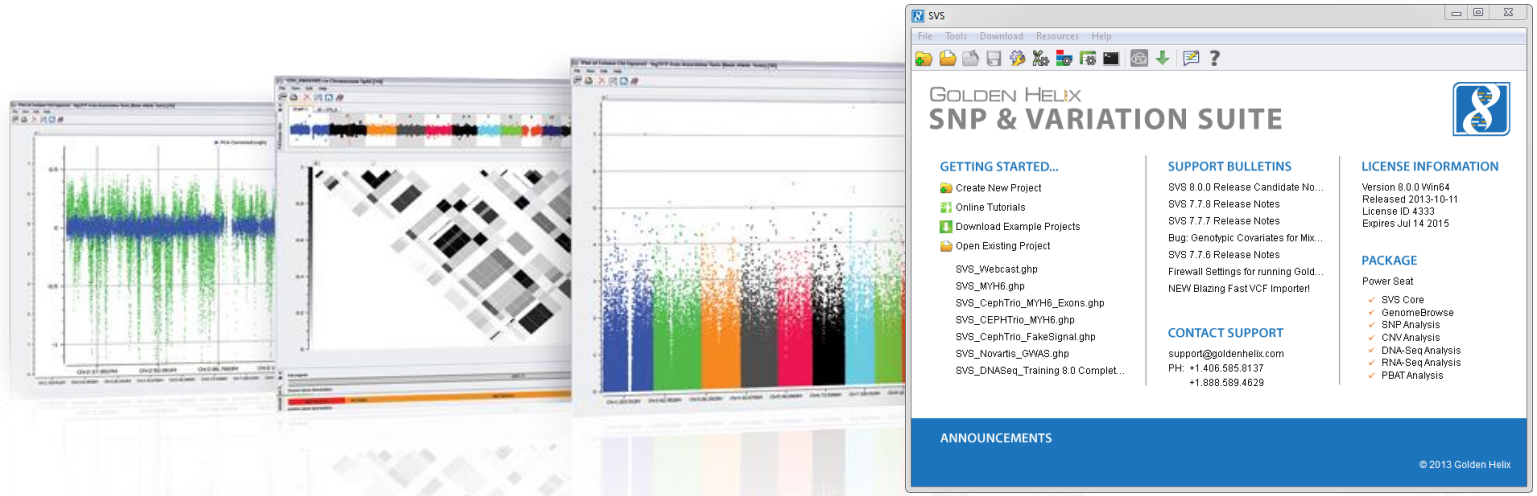
Use the Questions pane in your GoToWebinar window



Agenda

1. Tips for DNAseq
2. Data Management Tricks
3. Build Custom Genome
4. Questions!

SNP & Variation Suite (SVS)



Core Features

- Powerful Data Management
- Rich Visualizations
- Robust Statistics
- Flexible
- Easy-to-use

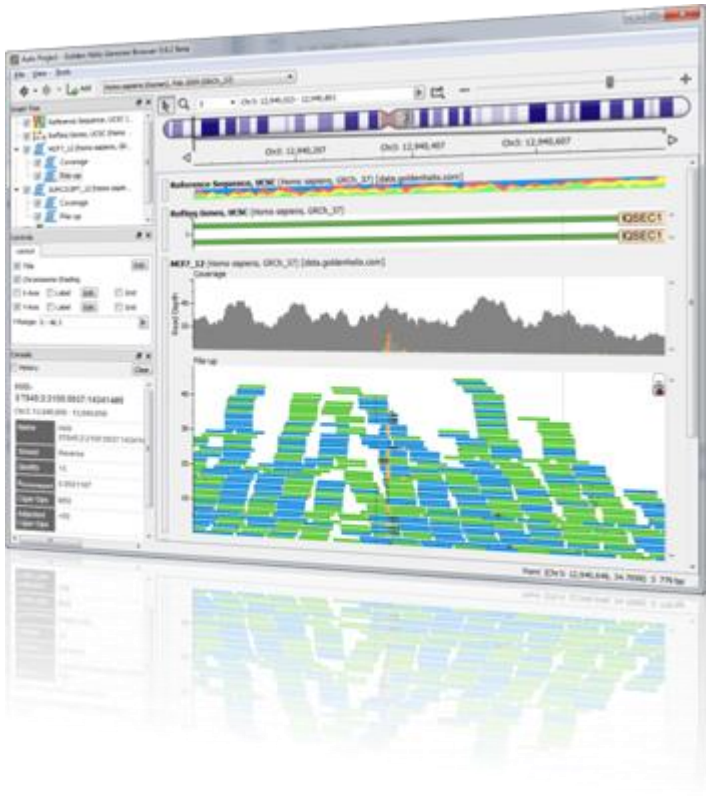
Applications

- Genotype Analysis
- DNA sequence analysis
- CNV Analysis
- RNA-seq differential expression
- Family Based Association

GenomeBrowse

- Free sequencing visualization tool
- Launched in 2011
- Makes the process of exploring DNA-seq and RNA-seq pile-up and coverage data intuitive and powerful
- Stream public annotations via the cloud
- Use it to validate variant calls, trio exploration, de Novo discovery, and more

Golden Helix GenomeBrowse™



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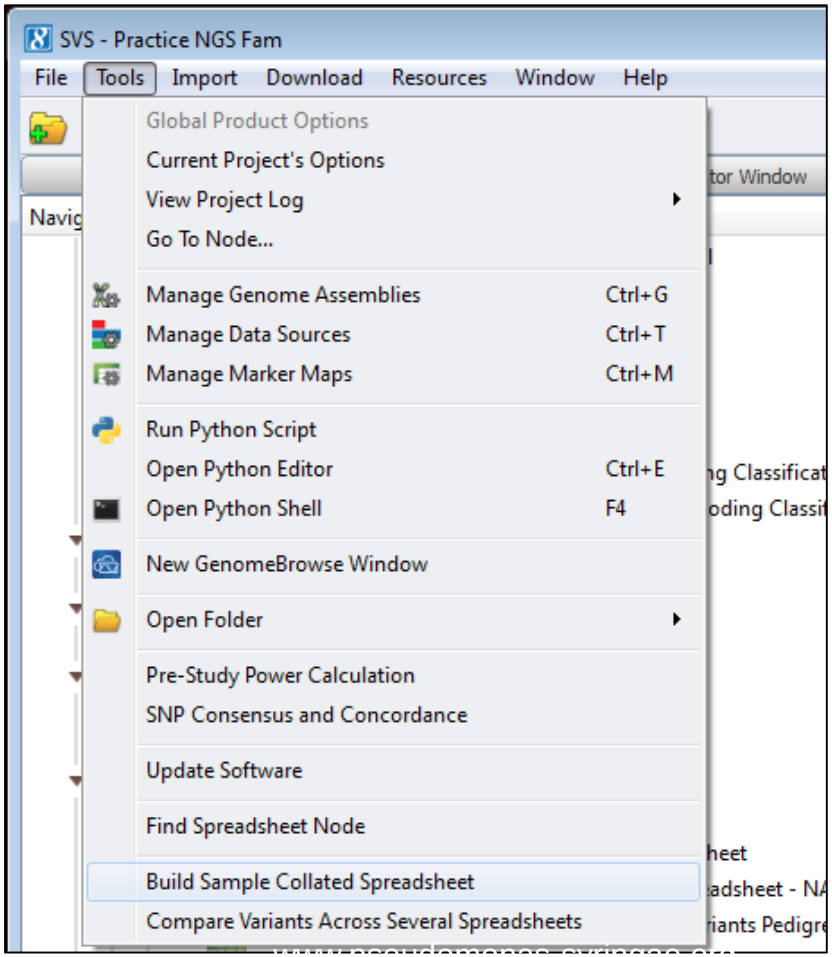
Tips for DNASeq

- Ability to create a collated and filtered spreadsheet from multiple inputs
  - Outputs data from where all spreadhseets overlap
- Sample use case – de Novo Variants
- Script: *Join or Merge Several Spreadsheets*



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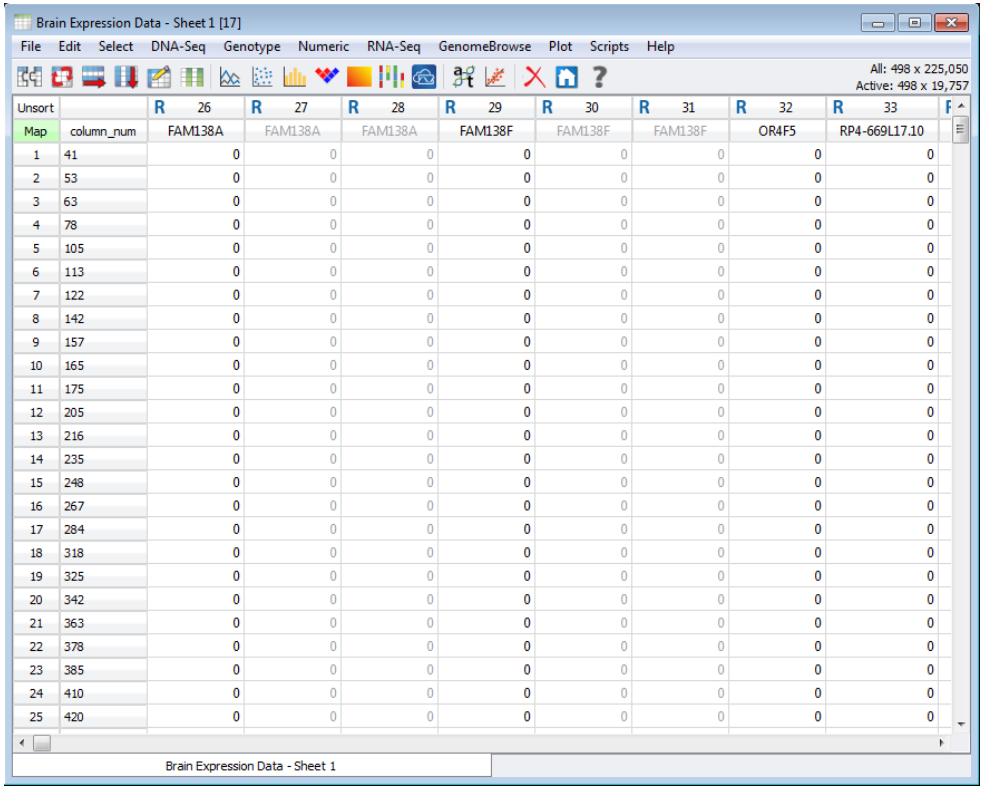
Data Management Tricks

- Activate by Gene List
- Activate by category in data columns
- Markers in LD block subset in Genome Browse
- Script: *Activate/Inactivate based on Genomic Position*
- Script: *Inactivate Duplicate Column Headers*



Unsort		R 26	R 27	R 28	R 29	R 30	R 31	R 32	R 33
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1	41	0	0	0	0	0	0	0	0
2	53	0	0	0	0	0	0	0	0
3	63	0	0	0	0	0	0	0	0
4	78	0	0	0	0	0	0	0	0
5	105	0	0	0	0	0	0	0	0
6	113	0	0	0	0	0	0	0	0
7	122	0	0	0	0	0	0	0	0
8	142	0	0	0	0	0	0	0	0
9	157	0	0	0	0	0	0	0	0
10	165	0	0	0	0	0	0	0	0
11	175	0	0	0	0	0	0	0	0
12	205	0	0	0	0	0	0	0	0
13	216	0	0	0	0	0	0	0	0
14	235	0	0	0	0	0	0	0	0
15	248	0	0	0	0	0	0	0	0
16	267	0	0	0	0	0	0	0	0
17	284	0	0	0	0	0	0	0	0
18	318	0	0	0	0	0	0	0	0
19	325	0	0	0	0	0	0	0	0
20	342	0	0	0	0	0	0	0	0
21	363	0	0	0	0	0	0	0	0
22	378	0	0	0	0	0	0	0	0
23	385	0	0	0	0	0	0	0	0
24	410	0	0	0	0	0	0	0	0
25	420	0	0	0	0	0	0	0	0

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SNP & VARIATION SUITE

[Demonstration]

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Build Custom genome

- Anopheles gambiae (Mosquito)
- Arabidopsis thaliana (Thale Cress)
- Bos taurus (Cow)
- Brassica rapa (Napa Cabbage)
- Caenorhabditis elegans (Round Worm)
- Canis familiaris (Dog)
- Capra hircus (Goat)
- Capsicum annuum (Hot Pepper)
- Carica papaya (Papaya)
- Citrullus lanatus (Watermelon)
- Cricetulus griseus (Chinese Hamster Ovary- CHO)
- Danio rerio (Zebrafish)
- Drosophila melanogaster (Fruit Fly)
- Equus caballus (Horse)
- Eucalyptus grandis (Eucalyptus)
- Felis catus (Cat)
- Gallus gallus (Chicken)
- Glycine max (Soybean)
- Gossypium raimondii (Cotton D)
- Heterocephalus glaber (Naked Mole-Rat)
- Leishmania infantum JPCM5 (Leishmania parasite)
- Macaca mulatta (Rhesus Monkey)
- Mus musculus (Mouse)
- Onchorhynchus mykiss (Rainbow Trout)
- Oryza sativa (Japanese Rice)
- Ovis aries (Sheep)
- Prunus persica (Peach)
- Rattus norvegicus (Norway Rat)
- Saccharomyces pombe (Baker's Yeast)
- Sorghum bicolor (Sorghum)
- Sus scrofa (Pig)
- Zea mays (Maize)

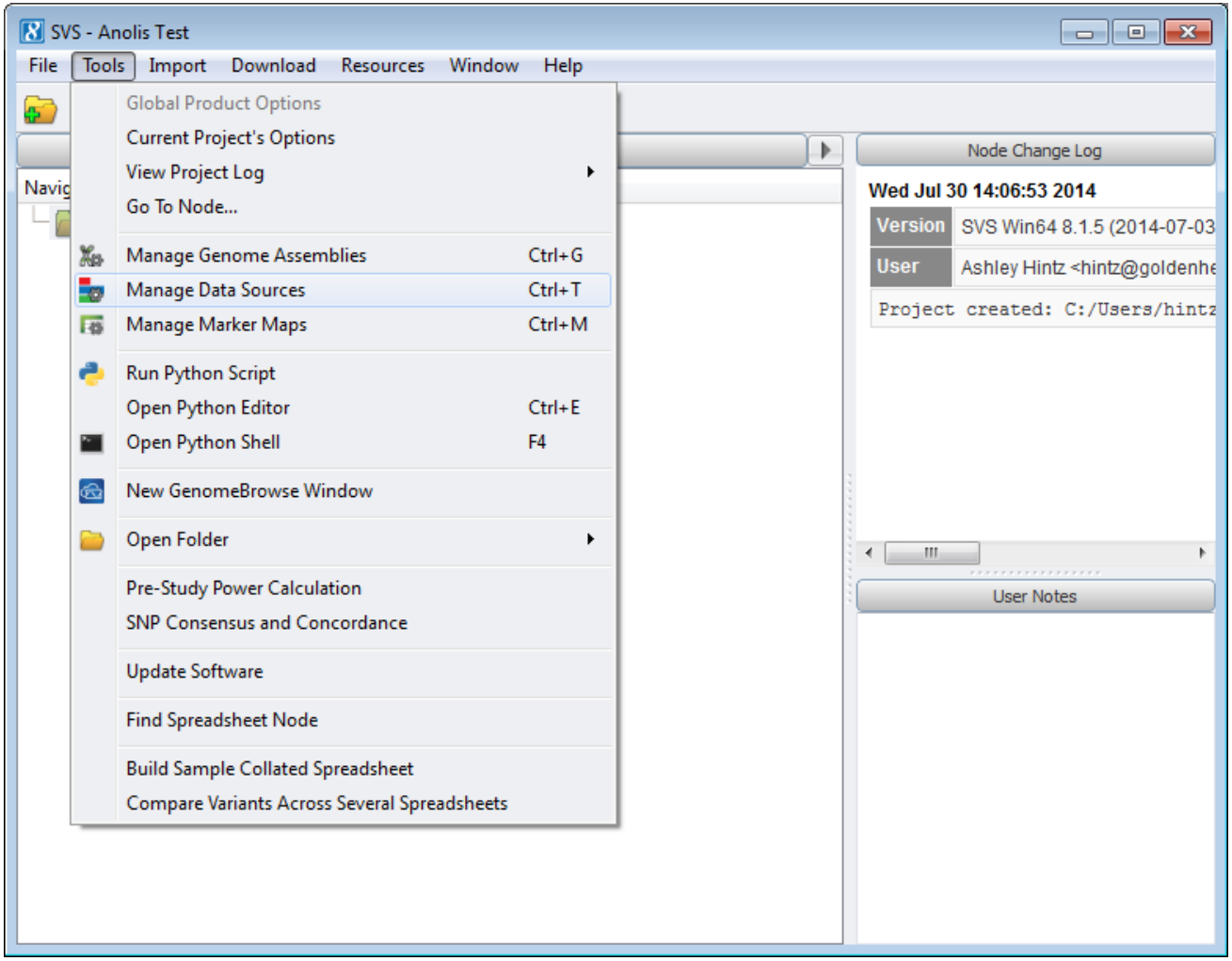
Build Custom Genome

- *Anolis carolinenesis* – Carolina Anole Lizard
- Downloaded from Ensembl
- Reference Genome
  - Separate FASTA file for each chromosome
- Gene Track
  - Single GTF file
- Originally published in Nature by Alföldi *et al.* (2011)
- 6 macro-chromosomes
  - Draft assembly 1.78GB
  - 17,472 protein-coding genes
  - 2,924 RNA genes

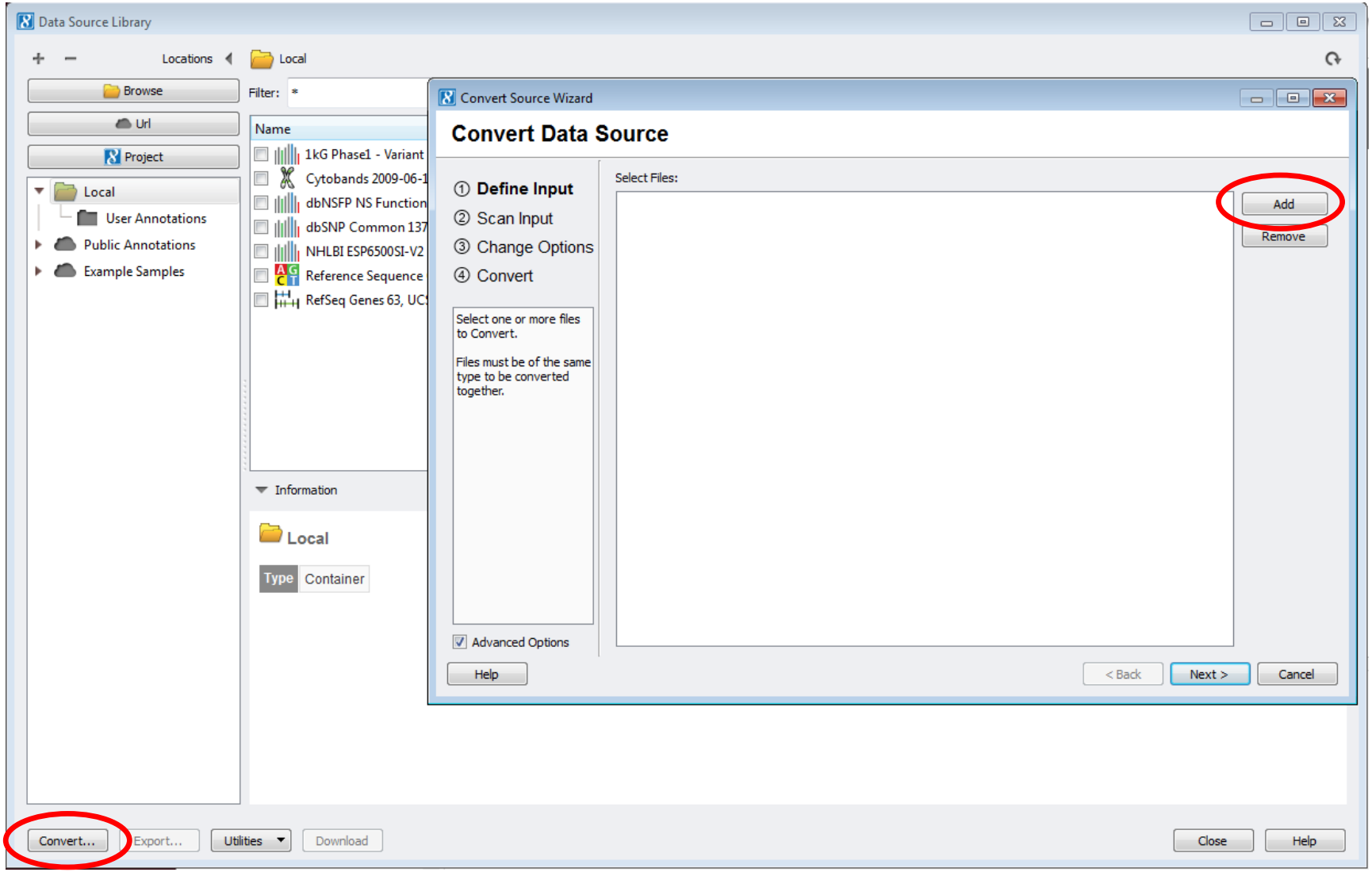
Photo courtesy R. Colin Blenis

Photo courtesy Robert Michniewicz

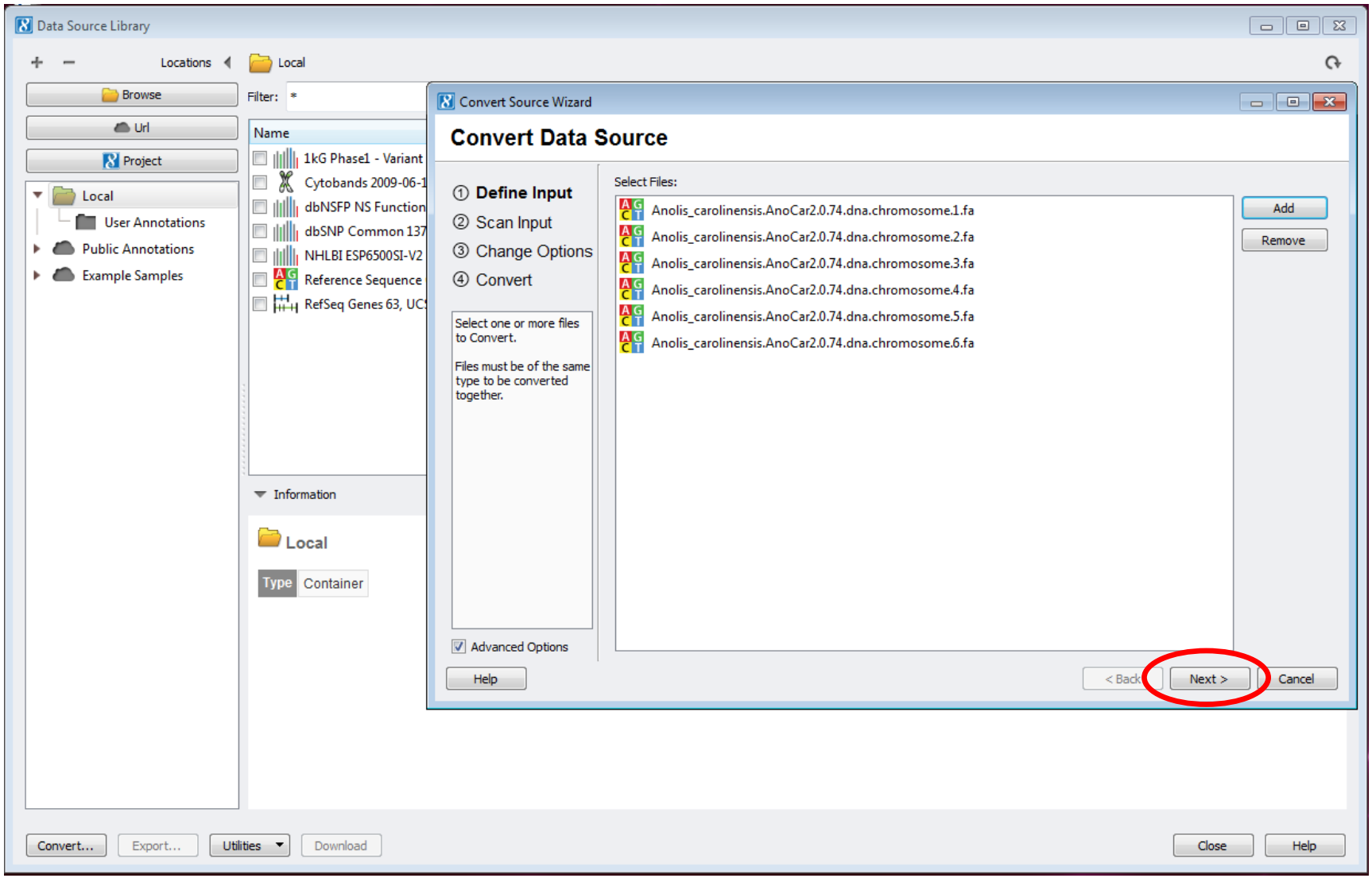
Build Custom Genome



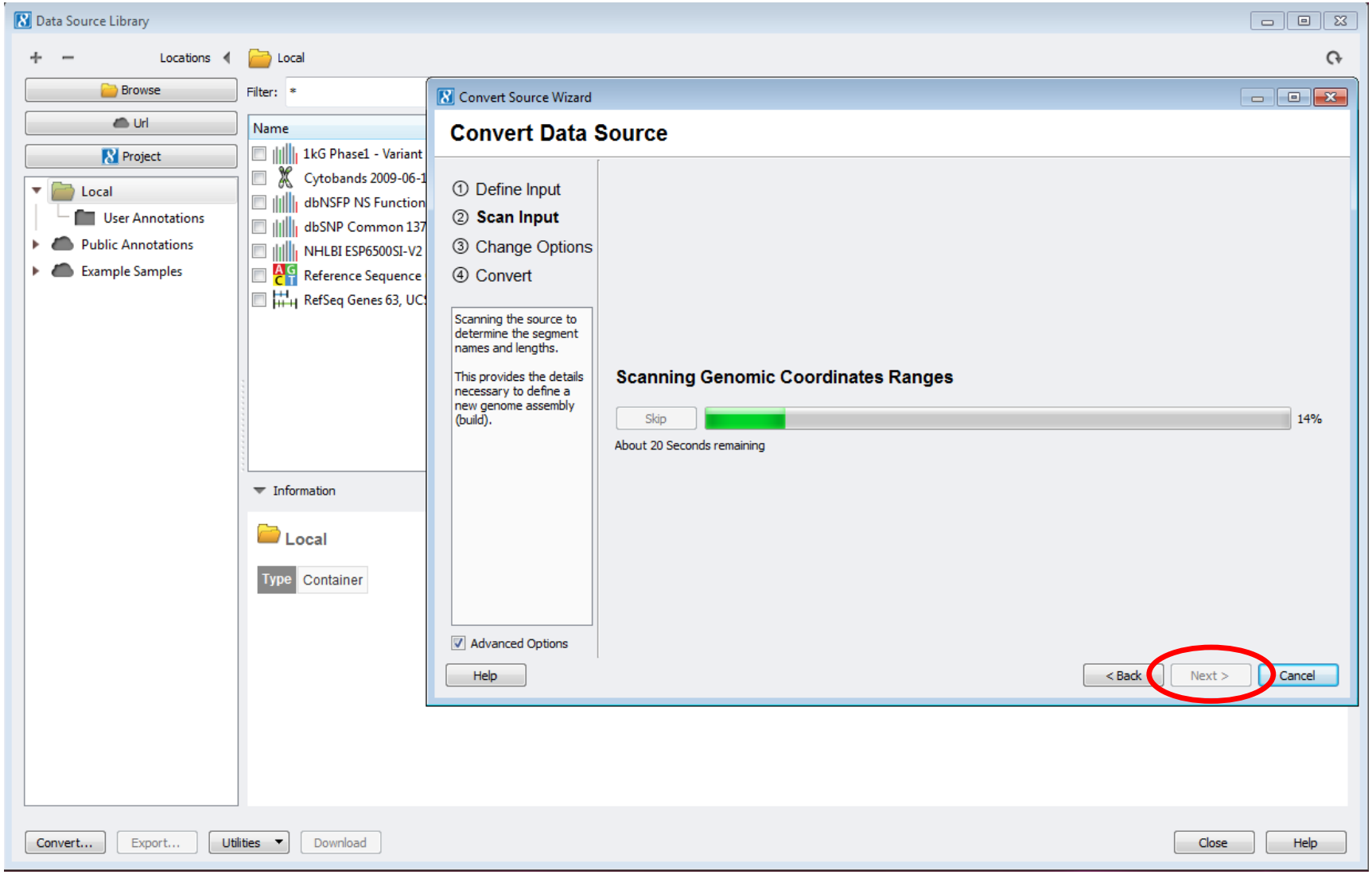
Build Custom Genome



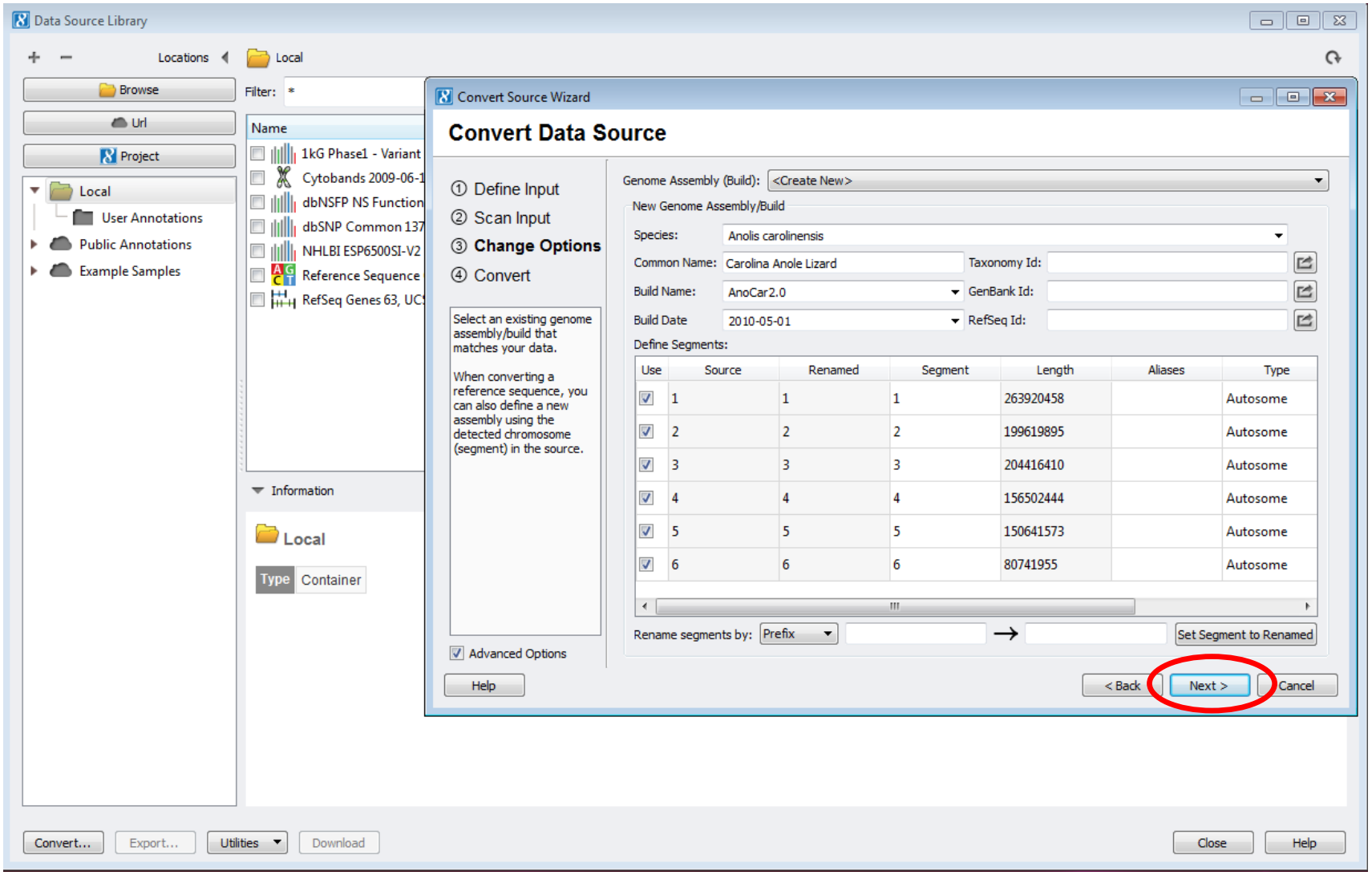
Build Custom Genome



Build Custom Genome



Build Custom Genome



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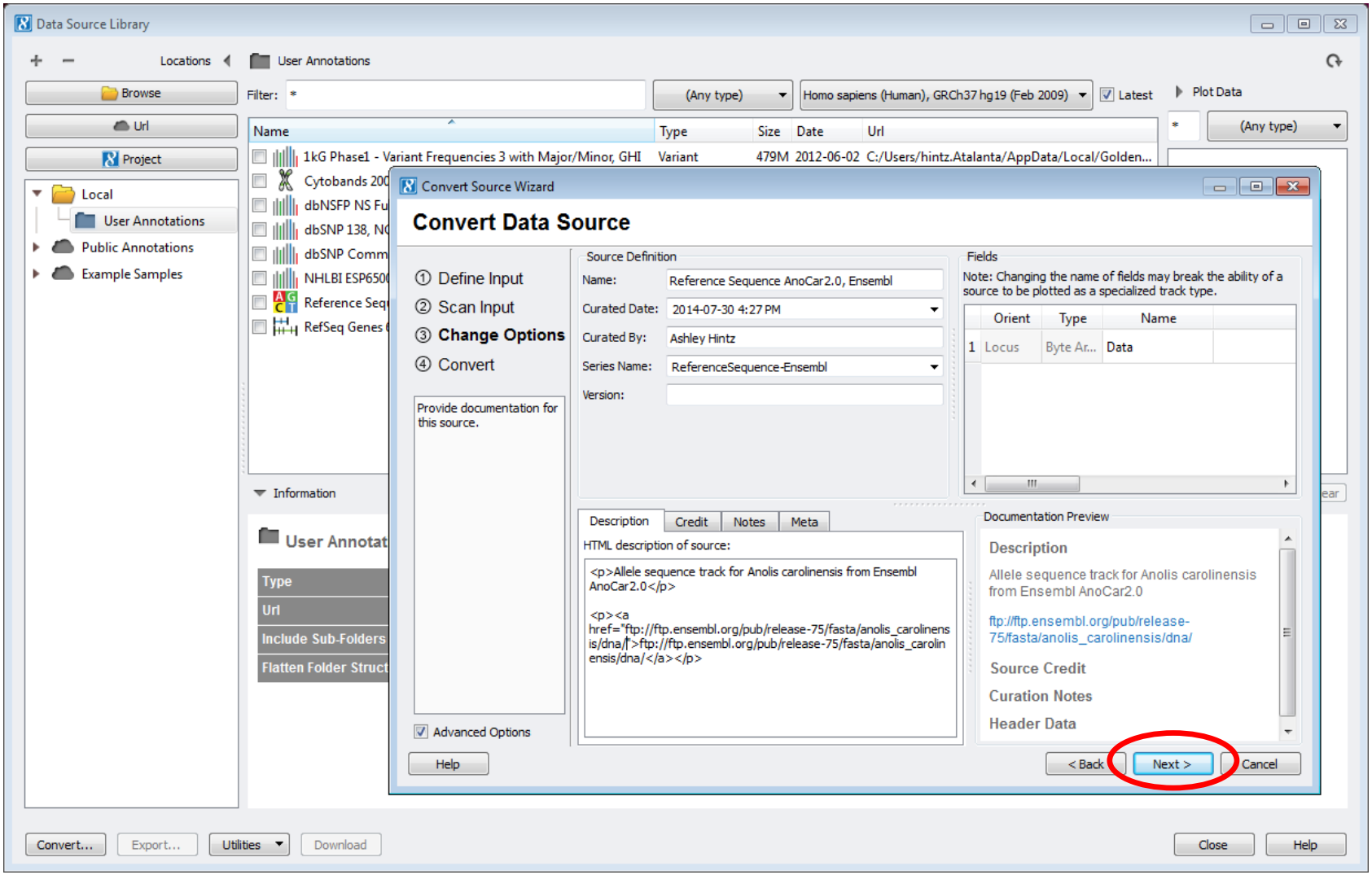

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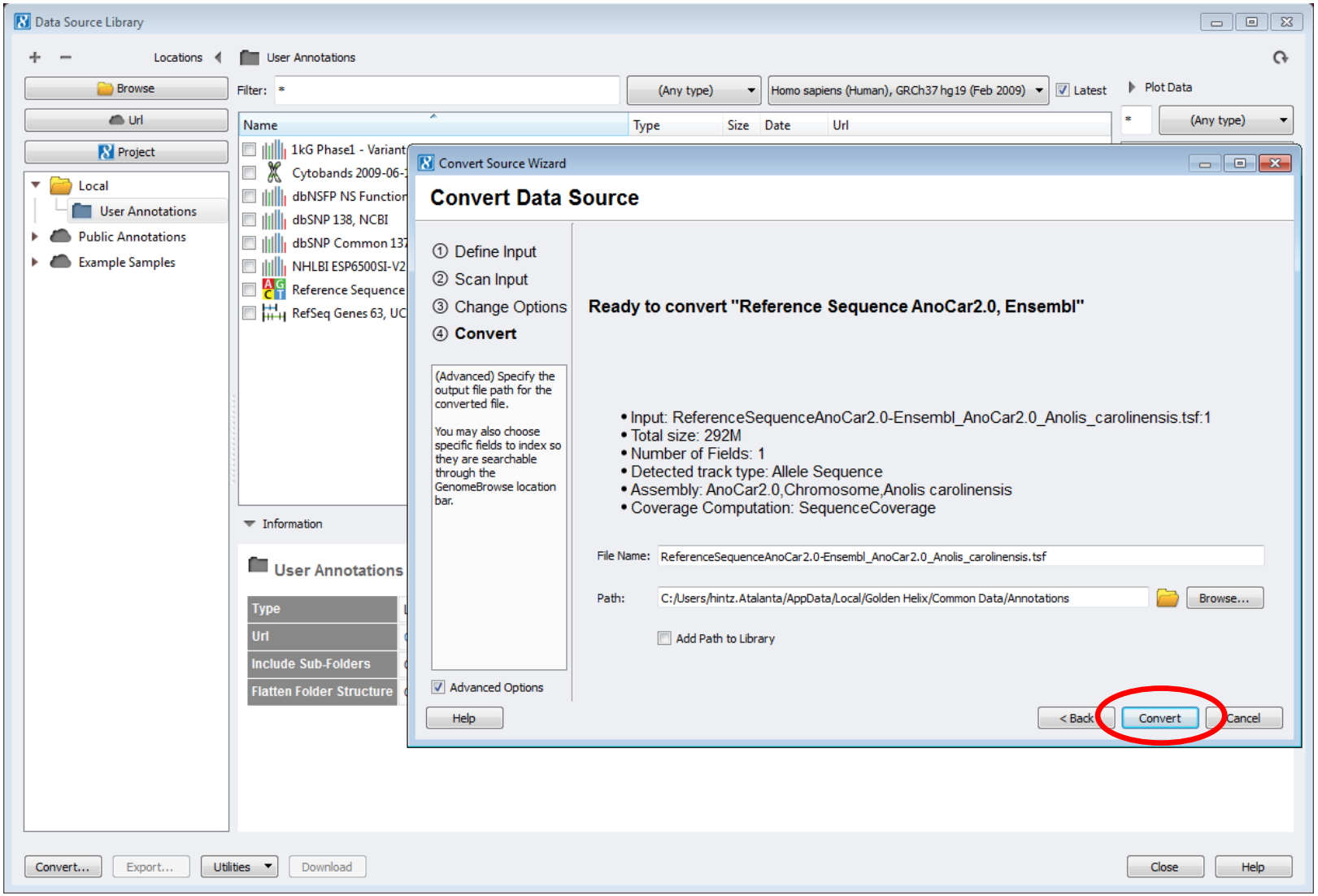

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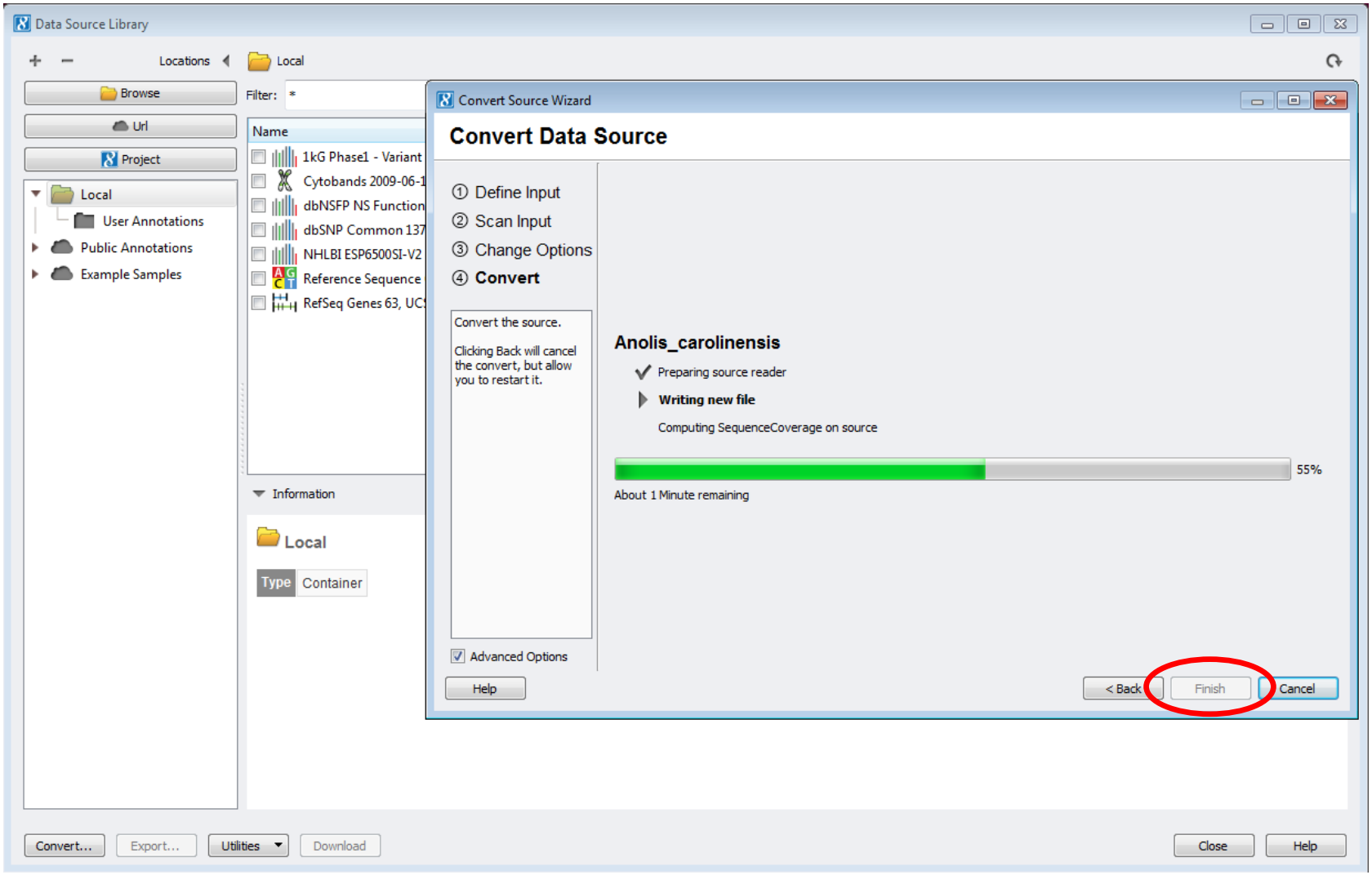

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[Demonstration]

Questions or more info:

- Email info@goldenhelix.com
- Request an evaluation of the software at www.goldenhelix.com