HapMap Concordance Check

for the Affymetrix® GenomeWideSNP_6 training kit

Genotyping Console 2.0 can calculate genotype concordance between your data and reference data. This document describes how you can check the concordance of the training kit samples against the HapMap reference genotypes originally obtained from www.hapmap.org.

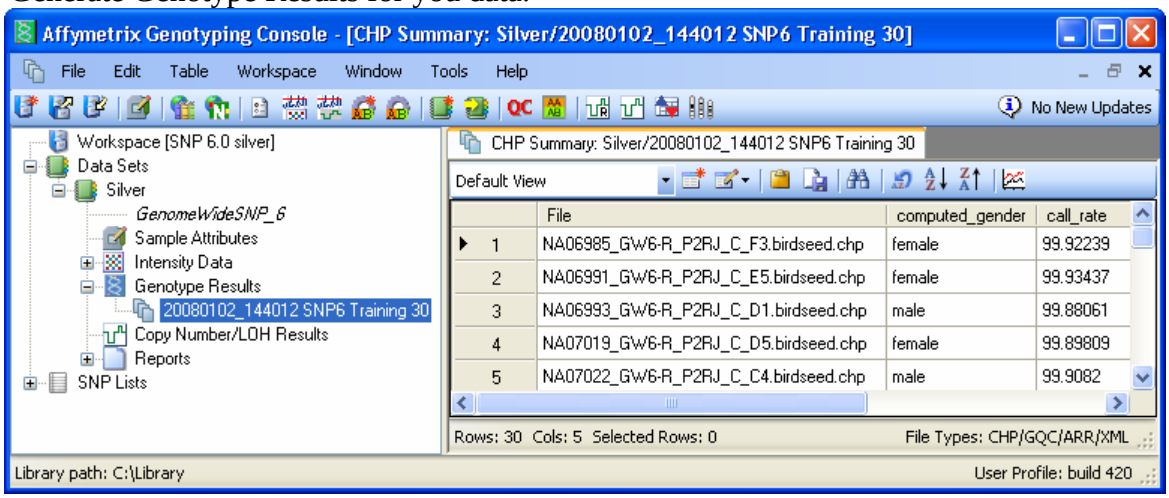
Genotyping Console 2.0 has an operation called "Run CHP vs. TXT Concordance Check". This operation compares multiple CHP files vs. a *single* reference sample's genotypes, stored in the TXT file. Since we want to compare CHP data vs. multiple samples' reference genotypes, we will instead use the operation "**Run CHP vs. CHP Concordance Check**". The supplied reference CHPs are synthetically generated, using data obtained from www.hapmap.org. The reference CHPs contain the subset of SNP_6 markers for which HapMap genotypes exist (HapMap data release 22). This amounts to 821,664 SNPs for each of the 30 unique HapMap samples in the training kit:

	Reference
1	SNP6_NA06985_HapMapReference.chp
2	SNP6_NA06991_HapMapReference.chp
3	SNP6_NA06993_HapMapReference.chp
4	SNP6_NA07019_HapMapReference.chp
5	SNP6_NA07022_HapMapReference.chp
6	SNP6_NA07056_HapMapReference.chp
7	SNP6_NA07345_HapMapReference.chp
8	SNP6_NA07348_HapMapReference.chp
9	SNP6_NA07357_HapMapReference.chp
10	SNP6_NA10831_HapMapReference.chp
11	SNP6_NA10835_HapMapReference.chp
12	SNP6_NA10846_HapMapReference.chp
13	SNP6_NA10855_HapMapReference.chp
14	SNP6_NA11831_HapMapReference.chp
15	SNP6_NA11832_HapMapReference.chp
16	SNP6_NA12144_HapMapReference.chp
17	SNP6_NA12145_HapMapReference.chp
18	SNP6_NA12155_HapMapReference.chp
19	SNP6_NA12156_HapMapReference.chp
20	SNP6_NA12248_HapMapReference.chp
21	SNP6_NA12249_HapMapReference.chp
22	SNP6_NA12740_HapMapReference.chp
23	SNP6_NA12750_HapMapReference.chp
24	SNP6_NA12751_HapMapReference.chp
25	SNP6_NA12753_HapMapReference.chp
26	SNP6_NA12762_HapMapReference.chp
27	SNP6_NA12763_HapMapReference.chp
28	SNP6_NA12801_HapMapReference.chp
29	SNP6_NA12812_HapMapReference.chp
30	SNP6_NA12813_HapMapReference.chp

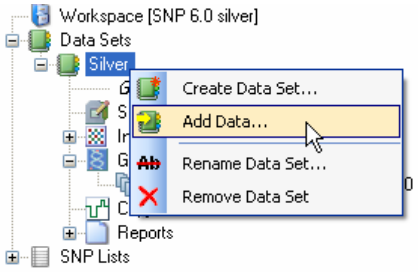
This procedure assumes you have some familiarity with Genotyping Console, and have already downloaded the HapMap reference CHP files.

Steps:

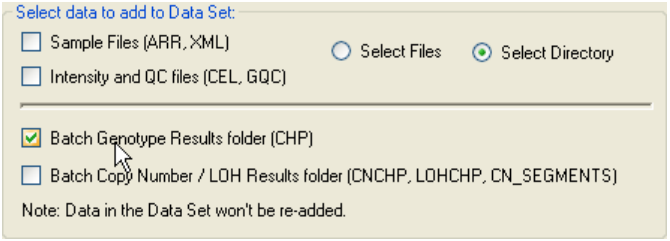
1. Generate Genotype Results for you data:



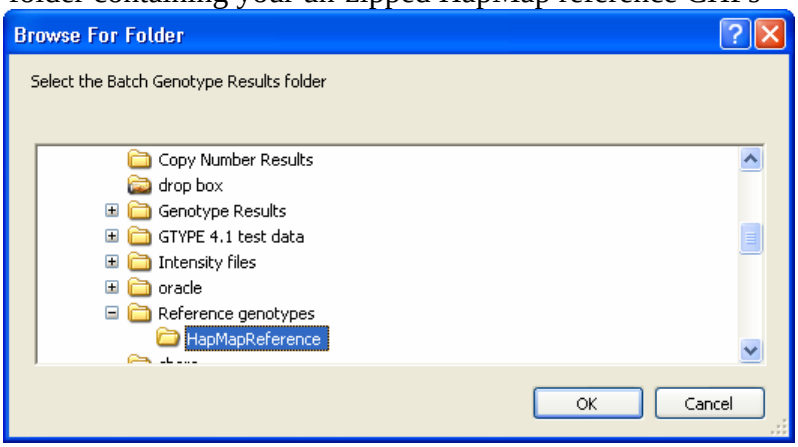
2. Now add reference data to this Data Set. Right-click on the same data set, and select "Add Data".



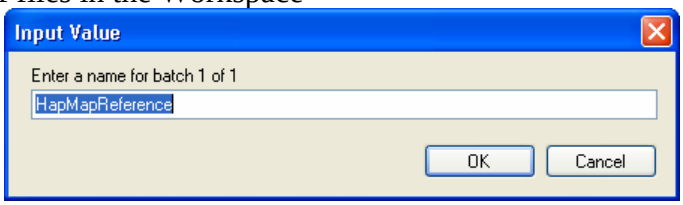
3. Check only the "Batch Genotype Results folder (CHP)" option in the data selection area. Click OK.



4. Browse to the folder containing your un-zipped HapMap reference CHPs

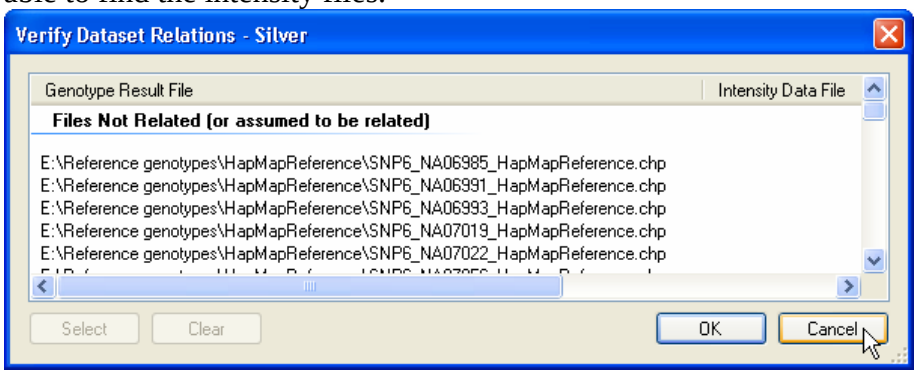


5. Name this batch of files in the Workspace

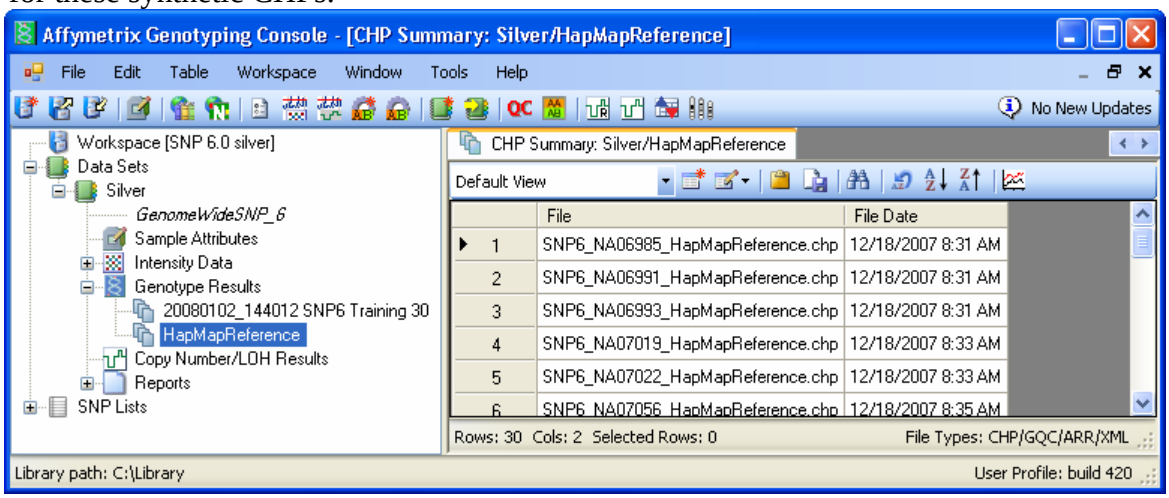


6. Because these are synthetic CHP files that weren't generated from Affymetrix CEL files, you will see the following message:

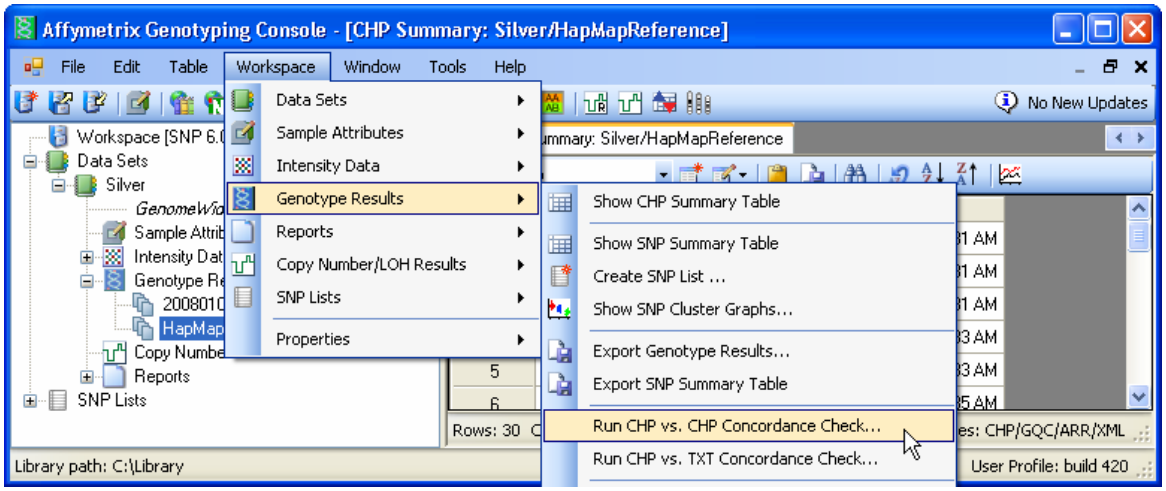
7. After clicking OK, you'll see the following window. You should click "Cancel," since you won't be able to find the intensity files.



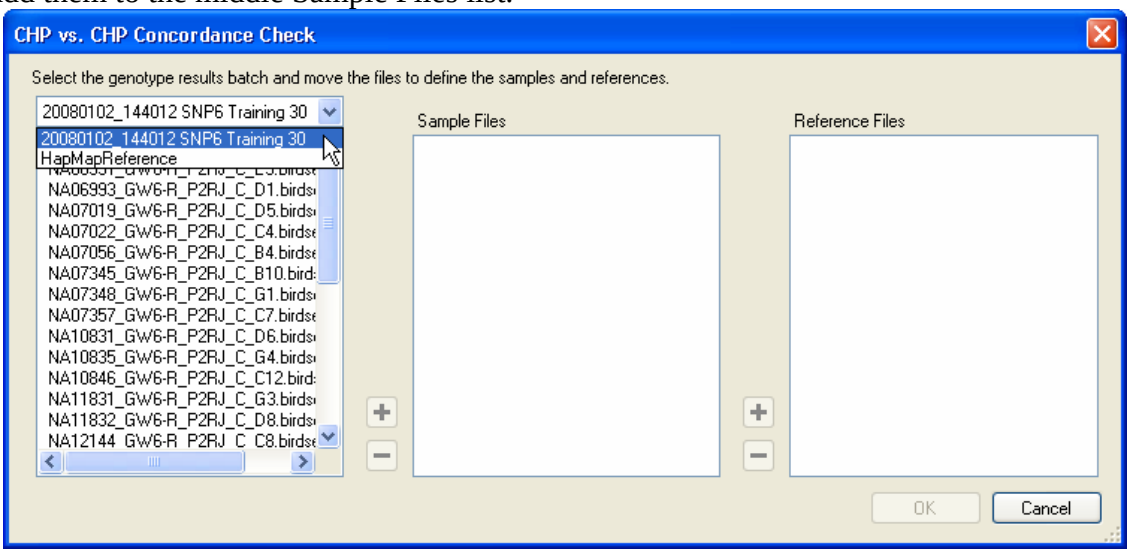
8. Your reference files will now be available in this Data Set. No summary statistics will be available in the CHP Summary table, nor will you be able to see SNP Cluster Graph signals for these synthetic CHPs:



9. From the Workspace menu, select Genotype Results > "Run CHP vs. CHP Concordance Check…"

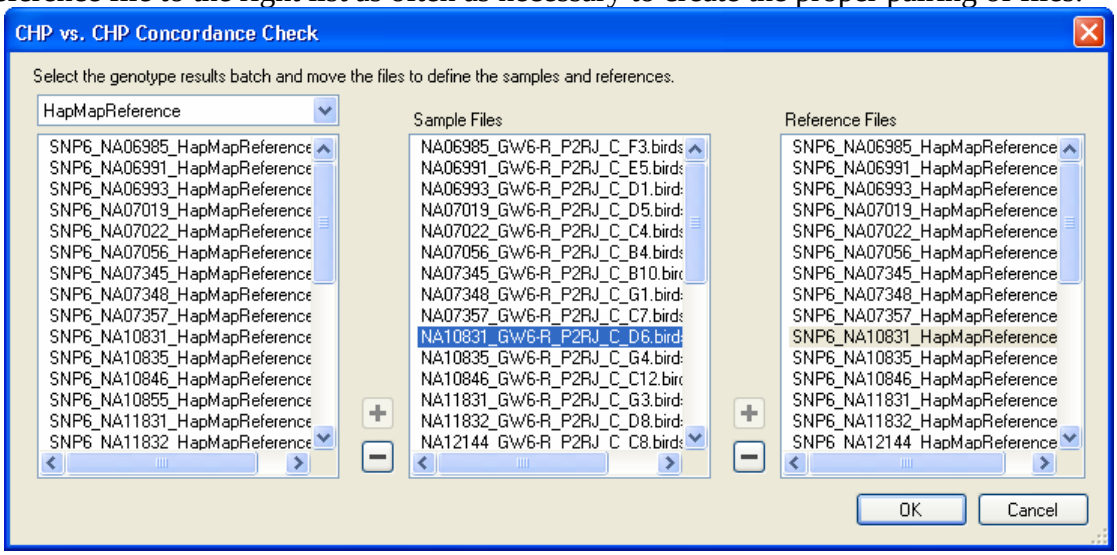


10. From the pull-down menu, select the test data to display in the left list. Then select some or all of the files in the list via appropriate control-click or shift-clicks, and select the + icon to add them to the middle Sample Files list.

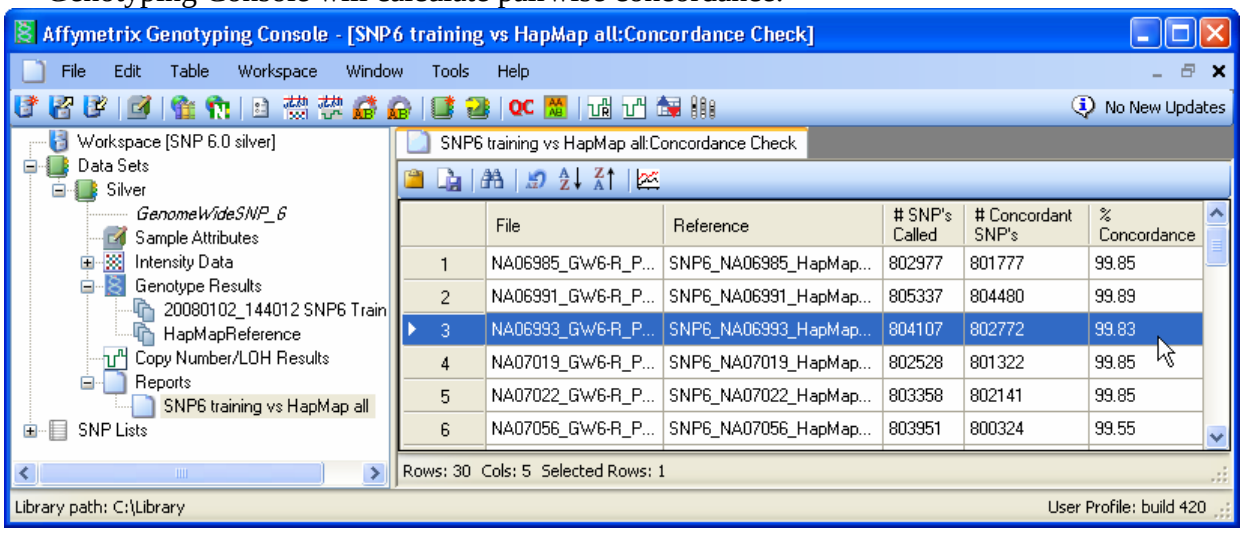


11. Do the same operation for the reference data, putting it into the right Reference Files list.

NOTE: This operation does pairwise comparison of files, based on order in the middle and right lists. For example, the file in row 10 of the middle list is compared against the row 10 file in the right list (see below). You may need to remove and re-add reference files to make sure they properly align with the sample files. If you have run the same sample repeatedly, you should add the reference file to the right list as often as necessary to create the proper pairing of files.



12. Click OK. You will be asked to save the concordance report file. After you do this, Genotyping Console will calculate pairwise concordance.



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