GAIN Genotyping Update

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GAIN Genotyping at Broad

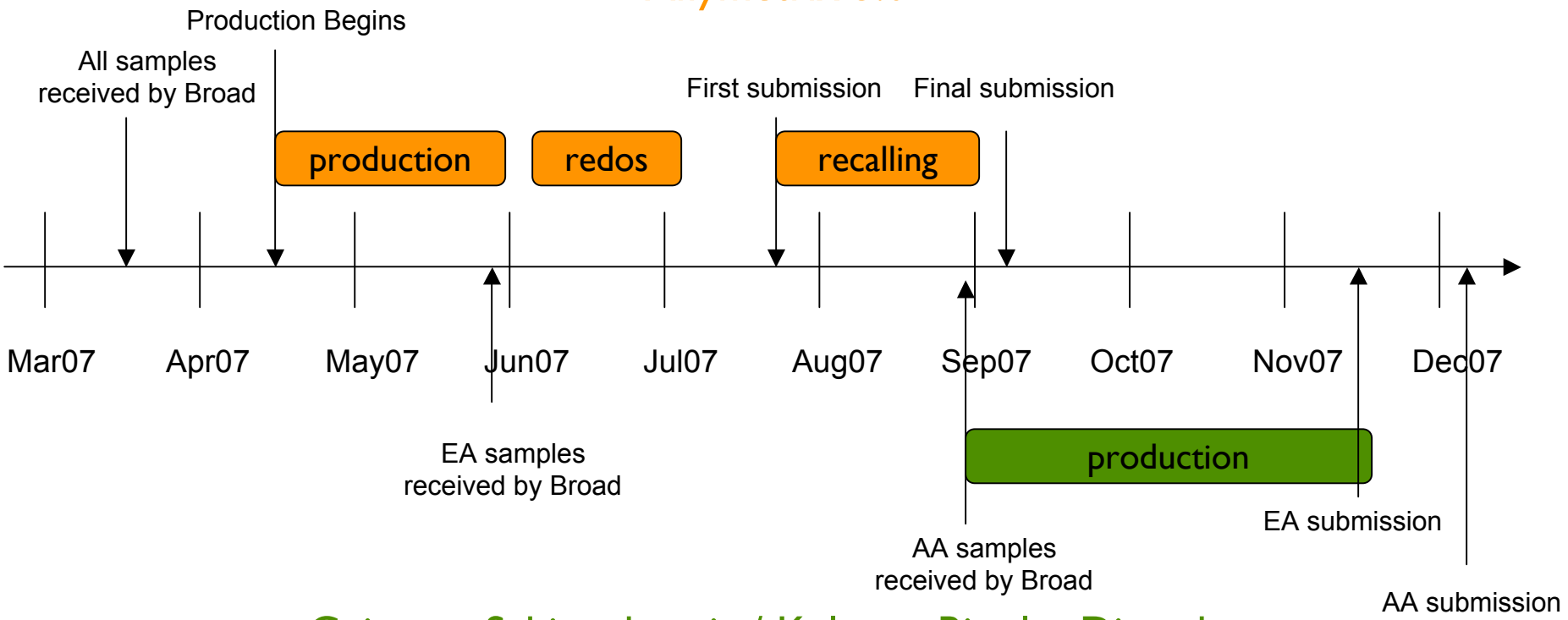
- Approximately 11,000 samples from three studies will be genotyped at Broad
- Affymetrix donated chips + kits
- Broad is supporting processing funds from:
  - Internal funds
  - NCRR genotyping grant subsidy program

GAIN Projects : Timeline

Warram study : Diabetic Nephropathy

3,118 samples

Affymetrix 5.0

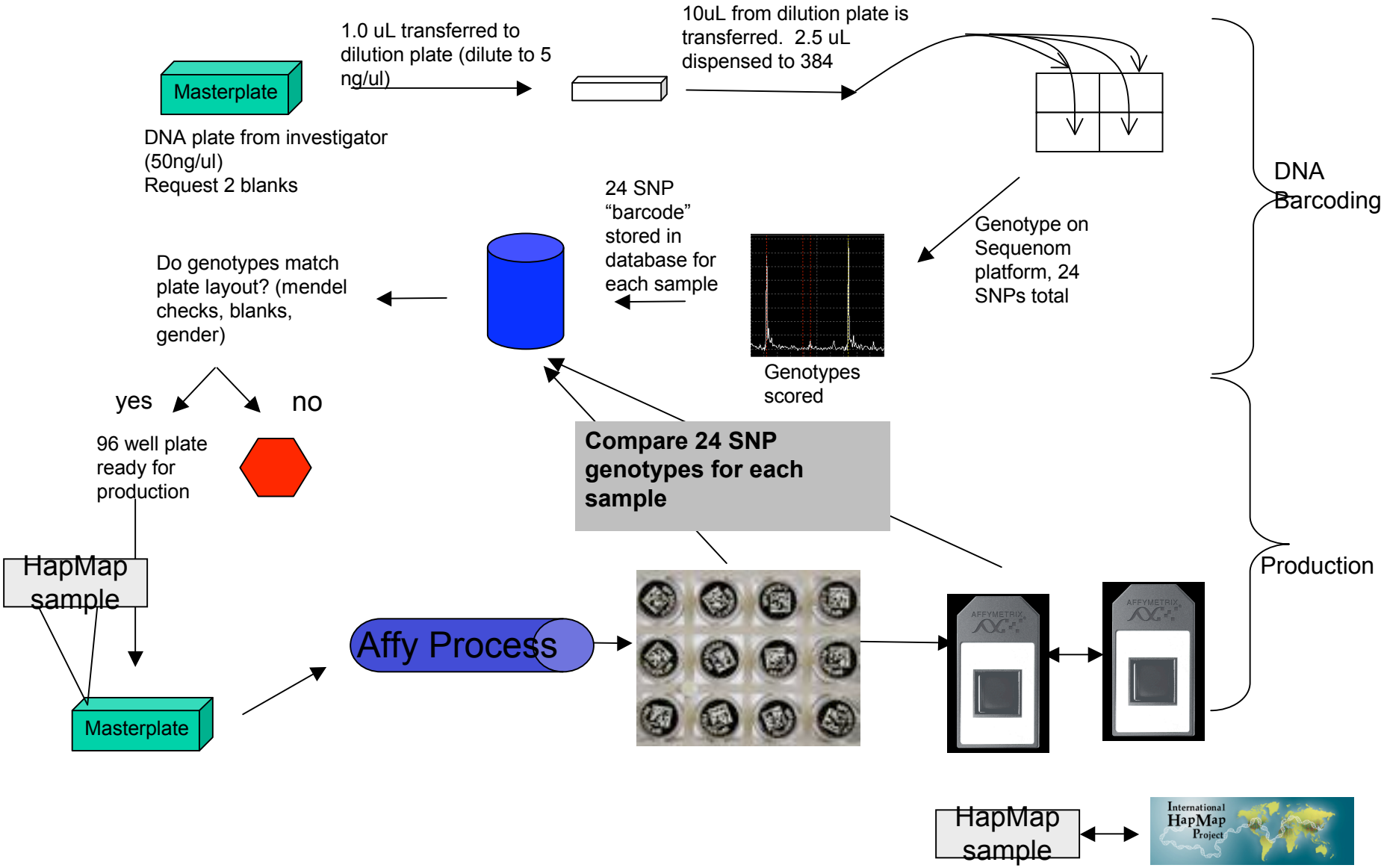


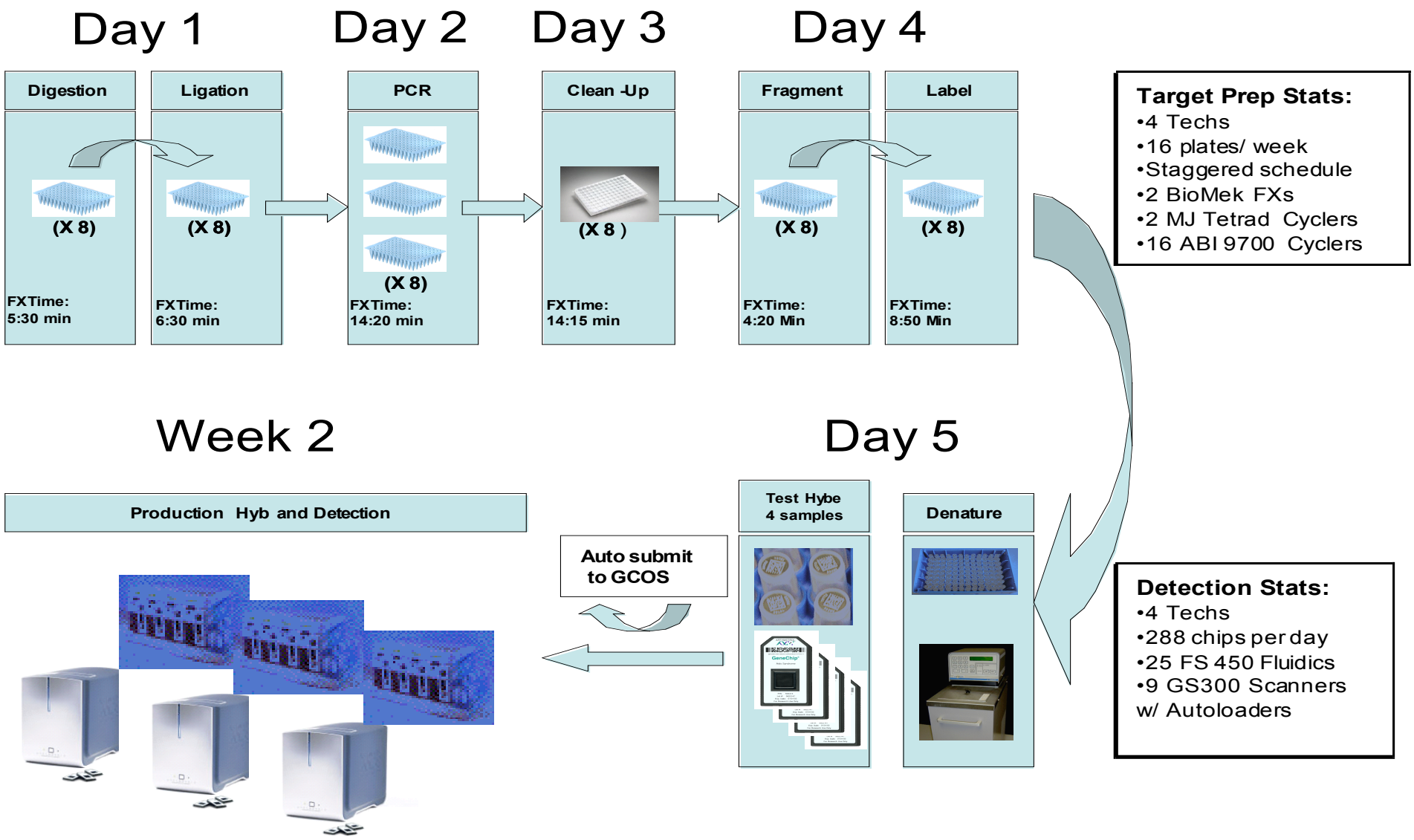
Gejman : Schizophrenia / Kelsoe : Bipolar Disorder

7,000 samples

Affymetrix 6.0

Workflow : sample tracking, QC




Day 1
Day 2
Day 3
Day 4
Digestion
Ligation
PCR
Clean -Up
Fragment
Label
(X 8)
(X 8)
(X 8)
(X 8)
(X 8)
(X 8)
FXTime:
5:30 min
FXTime:
6:30 min
FXTime:
14:20 min
FXTime:
14:15 min
FXTime:
4:20 Min
FXTime:
8:50 Min
Target Prep Stats:
•4 Techs
•16 plates/ week
•Staggered schedule
•2 BioMek FXs
•2 MJ Tetrad Cyclers
•16 ABI 9700 Cyclers
Week 2
Day 5
Production Hyb and Detection
Auto submit
to GCOS
Test Hybe
4 samples
Denature
Detection Stats:
•4 Techs
•288 chips per day
•25 FS 450 Fluidics
•9 GS300 Scanners
w/ Autoloaders

Data Flow and Filtering

Re-queue 1X

No

FQC pass?
FQC = 3,000 "fragile SNPs" called by DM algorithm

Is Fingerprint match >90%?

HapMap Concordance (1 / plate)

No

YES

Does plate have >75% passing chips ?

YES

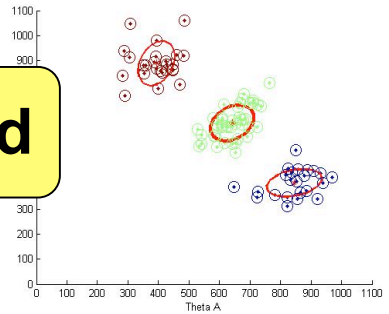
Is Call Rate>95%?

YES

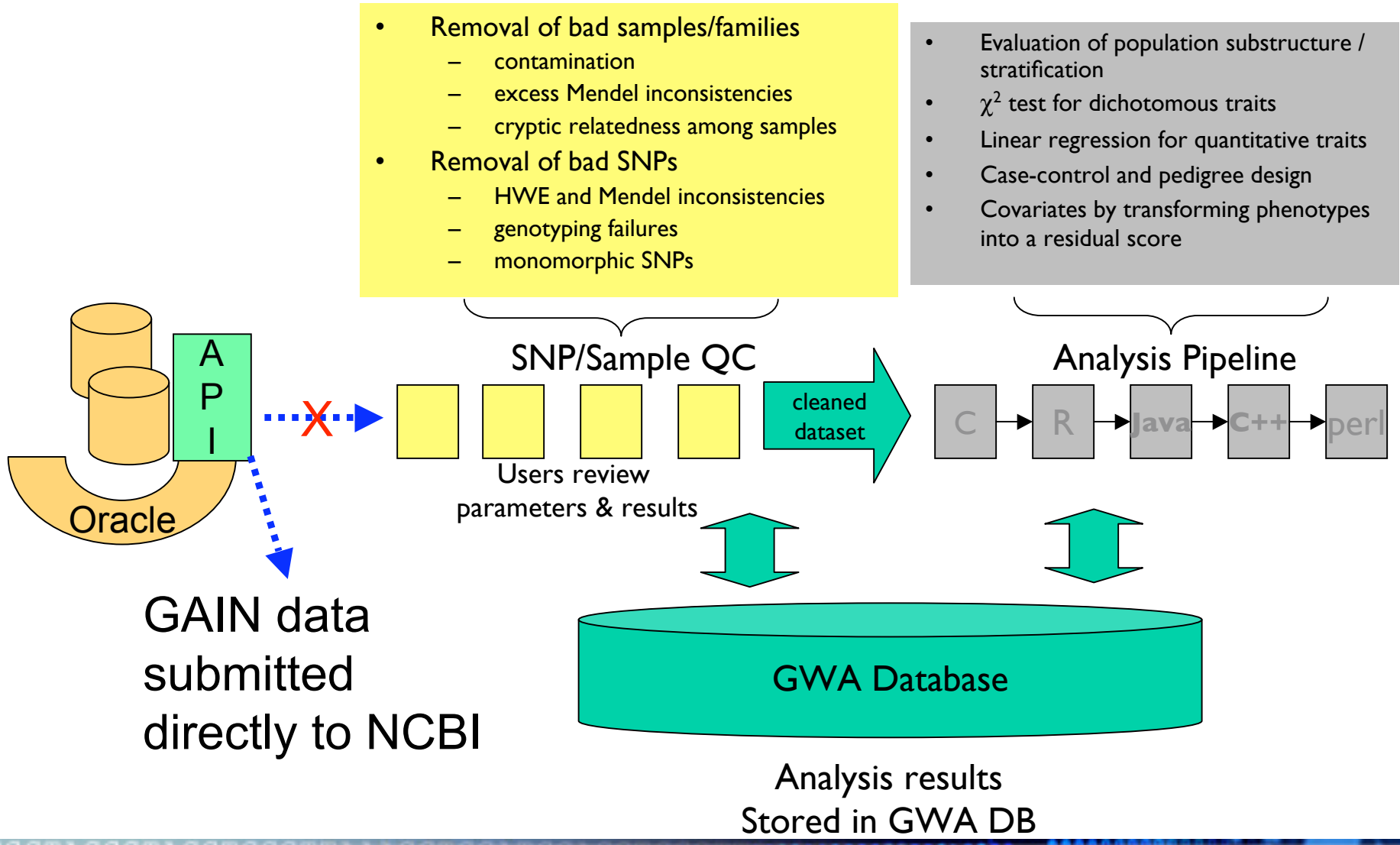
A P I

Oracle

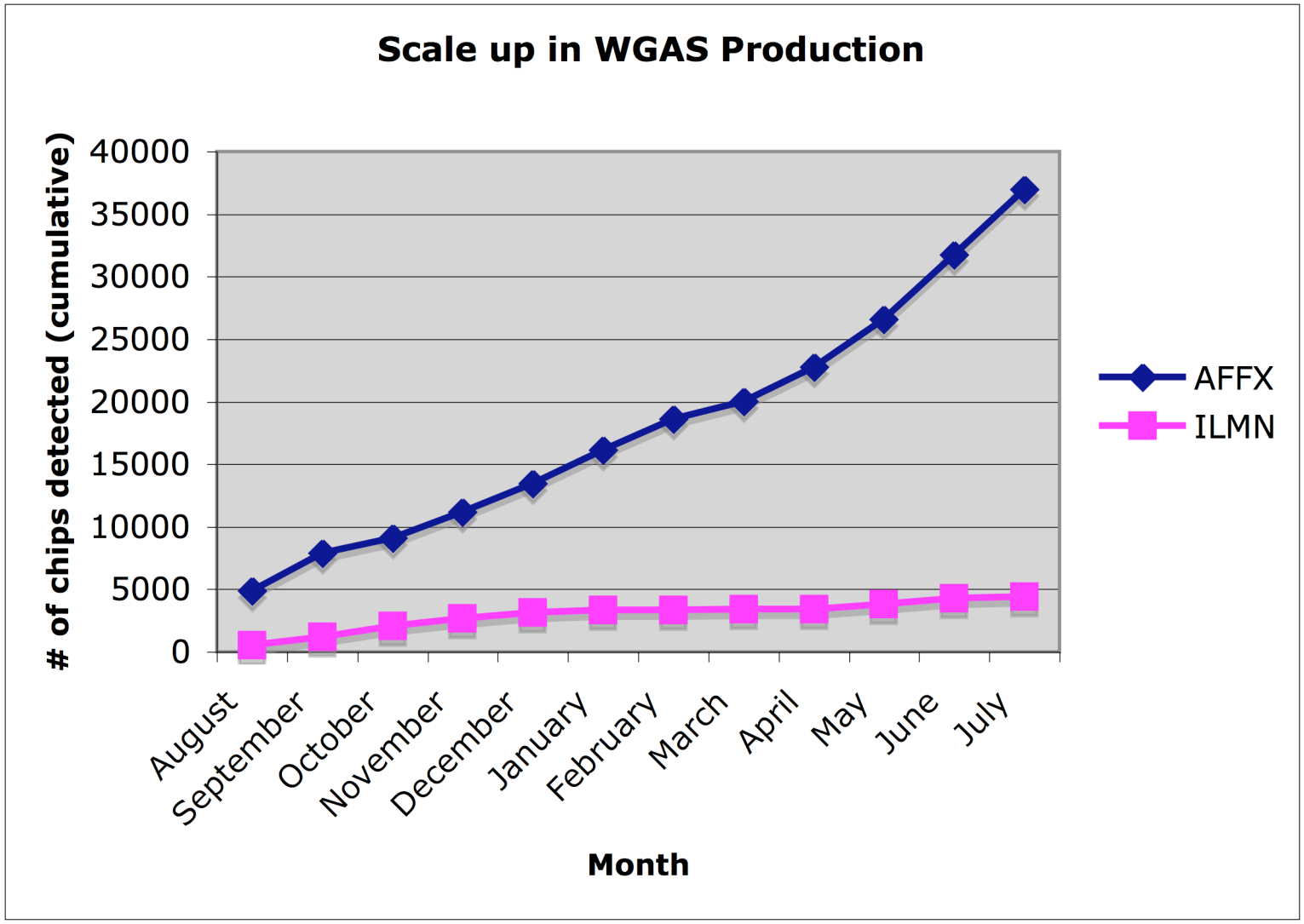
Birdseed



Broad Data Analysis Pipeline



Production Scale Up



Completed GWAS Studies

Genome-wide Association Studies Complete

Study	Multiple Sclerosis	Type II Diabetes	Bipolar Disorder	Lupus	AML	Autism	Diabetic Nephropathy	Schizophrenia	Myocardial Infarction
Investigators	IMSGC	DGI	Sklar/Scholnik	SLEGEN	Brown (MGH)	Autism Consortium	James Warram (Joslin)	Sklar/Sckolnick	MIGen
Platform	Affy 500K	Affy 500K	Affy 500K	ILMN 300	ILMN 300	Affy 5.0	Affy 5.0	Affy 6.0	Affy 6.0
Samples Submitted	1545	3313	4342	1165	2003	3166	3118	4820	5240
Samples Successful	1510 (98%)	3160 (95%)	4171 (96%)	941 (95%)	1760 (97%)	3103 (98%)	2985 (96%)	pending	5082 (97%)

30,000 samples scanned
~40,000 samples queued up