



CynerGene LLC

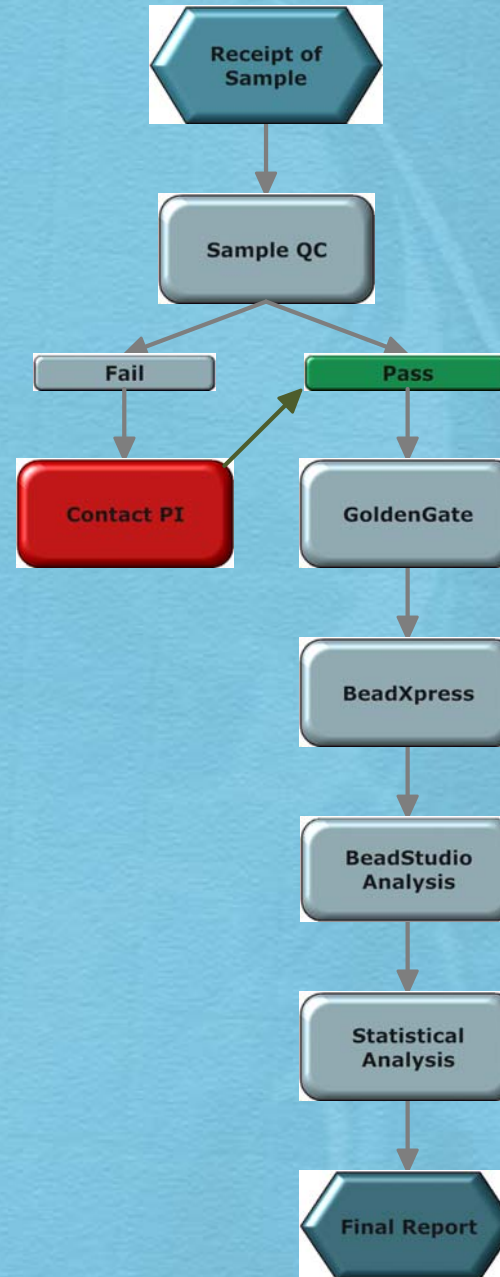
Multiple Approaches to Focused
SNP Genotyping using Illumina's
BeadXpress

Background...

- 2 Years Established
- GLP Compliant
- Genomic Service providers
 - Gene Expression
 - DNA Methylation
 - Focused SNP Genotyping***
 - Sample Handling, Extraction ...



Pipeline...

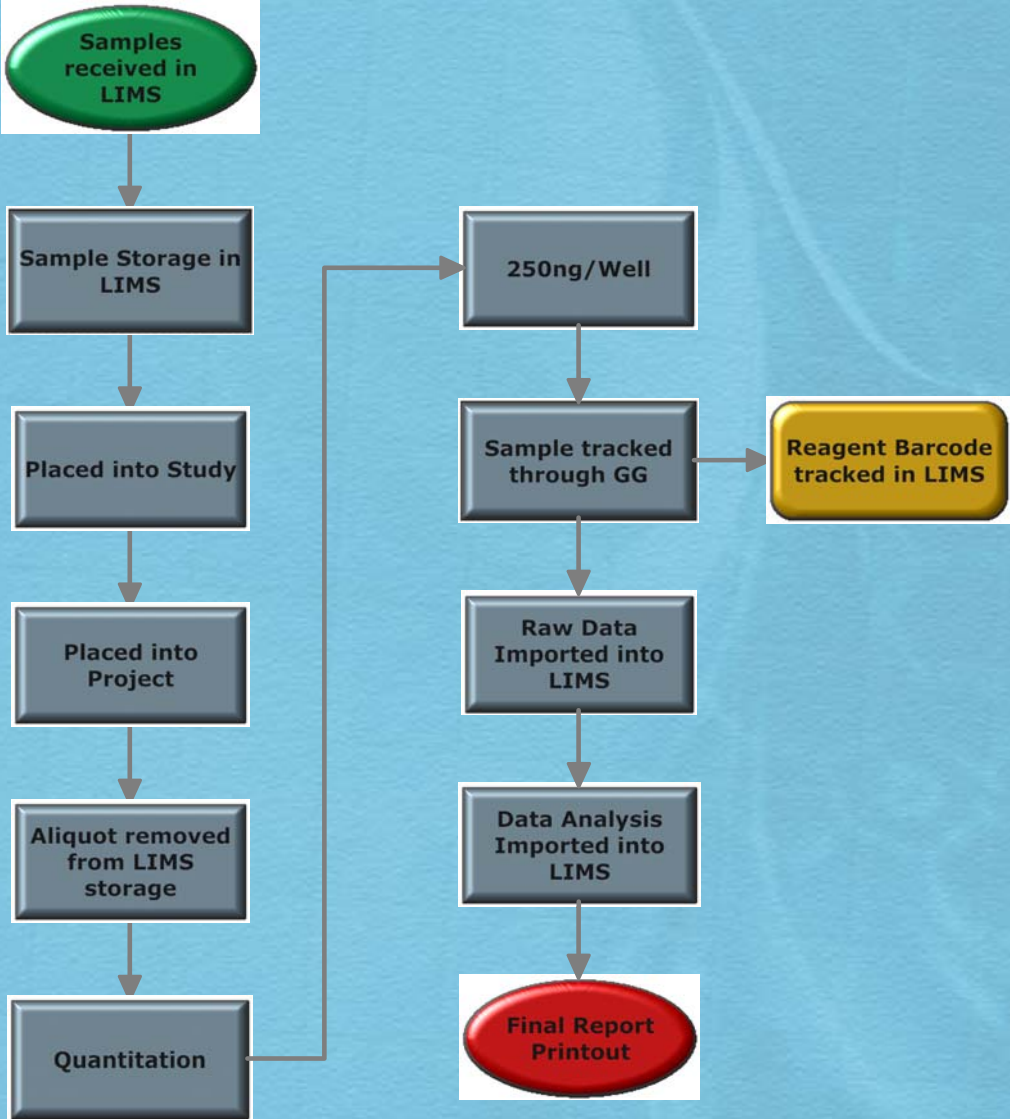




Sample QC

- Different Extraction Methods
 - Blood
 - Saliva
 - Buccal Swab
- 250ng/sample/well
 - 300ng/sample
- Samples Identified and Received in LIMS
- Quantitation
- Gel Electrophoresis

LIMS

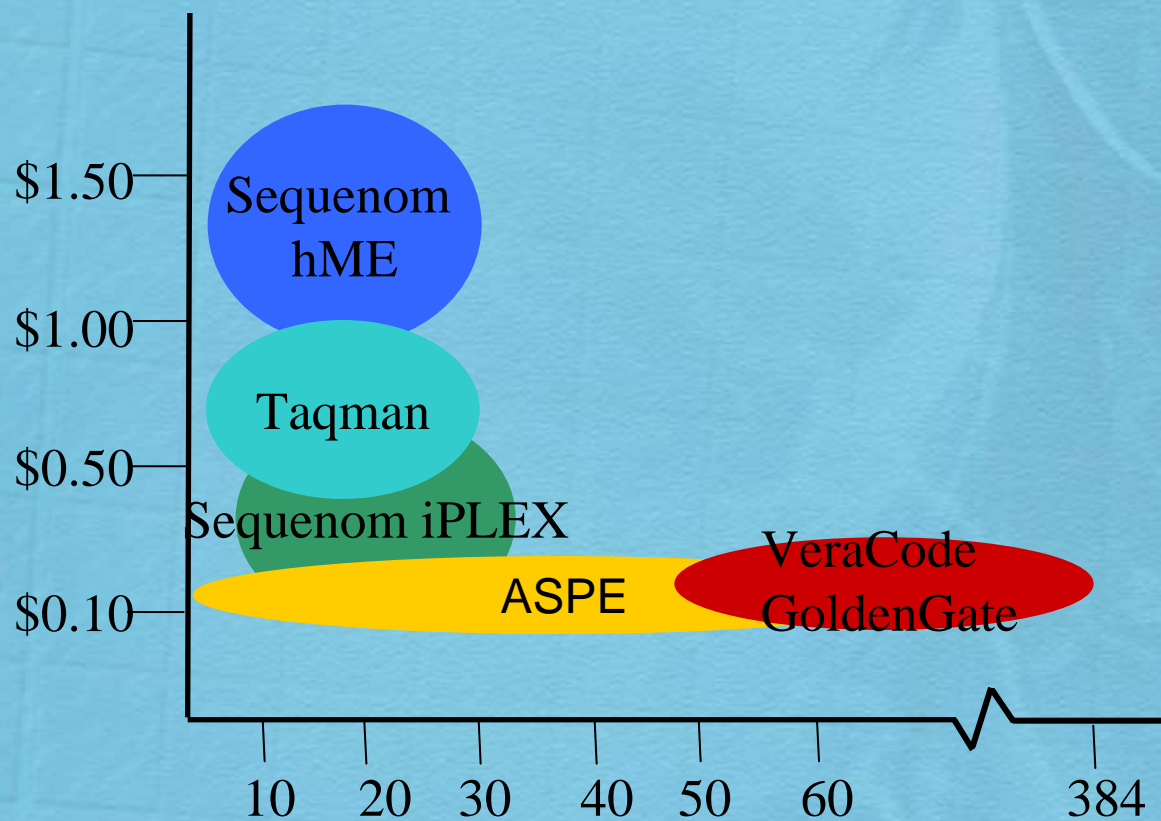




Benefits of BeadXpress

- Easy to follow protocols
- Robust Assay
- Reliable data
- Throughput:
 - 4 plates/technician (1.5 days)
- Cost effective
 - Decreased sample usage
 - Increased SNP number

Cost Analysis





Genotyping Application...

- Experiment 1
 - SNP Scoring
 - Sample Volume
- Experiment 2
 - Sample Volume Cont'd
 - Species Differentiation

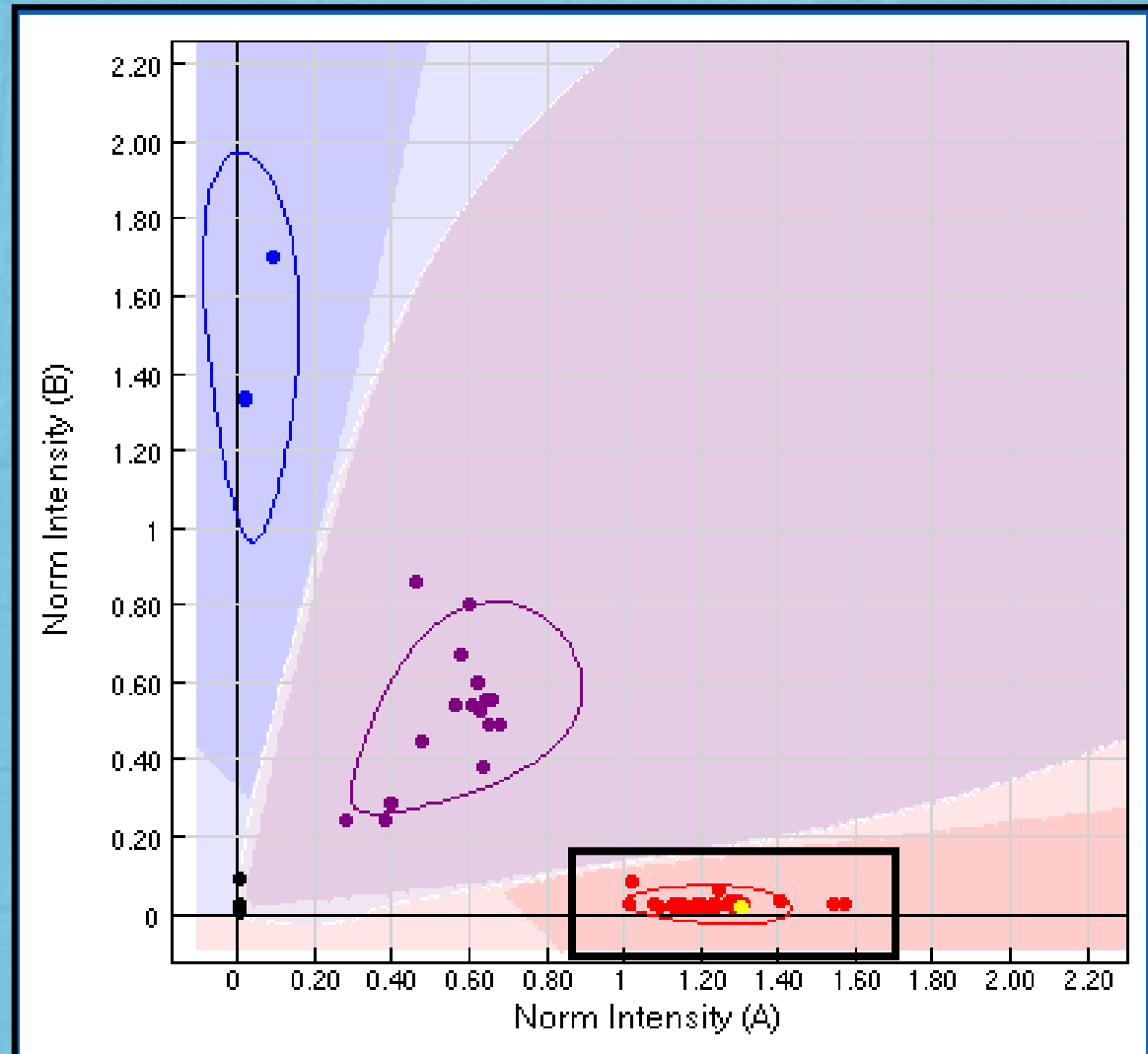


Experiment 1

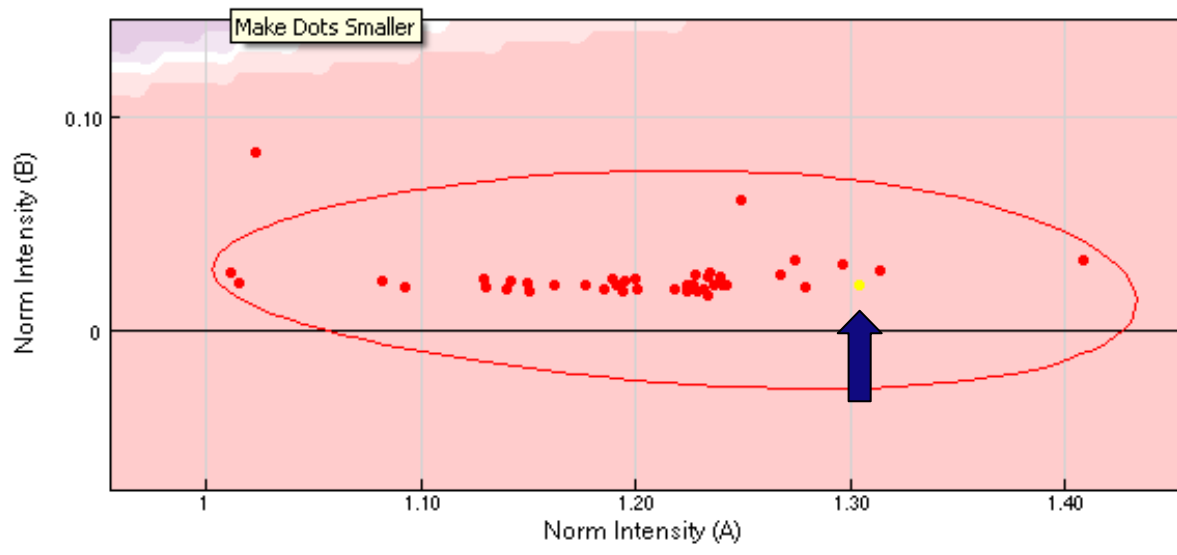
- 384 SNPs
- Overall Mean Call Rate per SNP 98.88%
- 9 SNPs <90%
 - Cut-off point 40 bp
- Low DNA amount

Score	Number SNPs
1.1	56
1.00-0.90	102
0.899-0.80	59
0.799-0.70	59
0.699-0.60	40
0.599-0.50	31
<0.499	37
Total	384

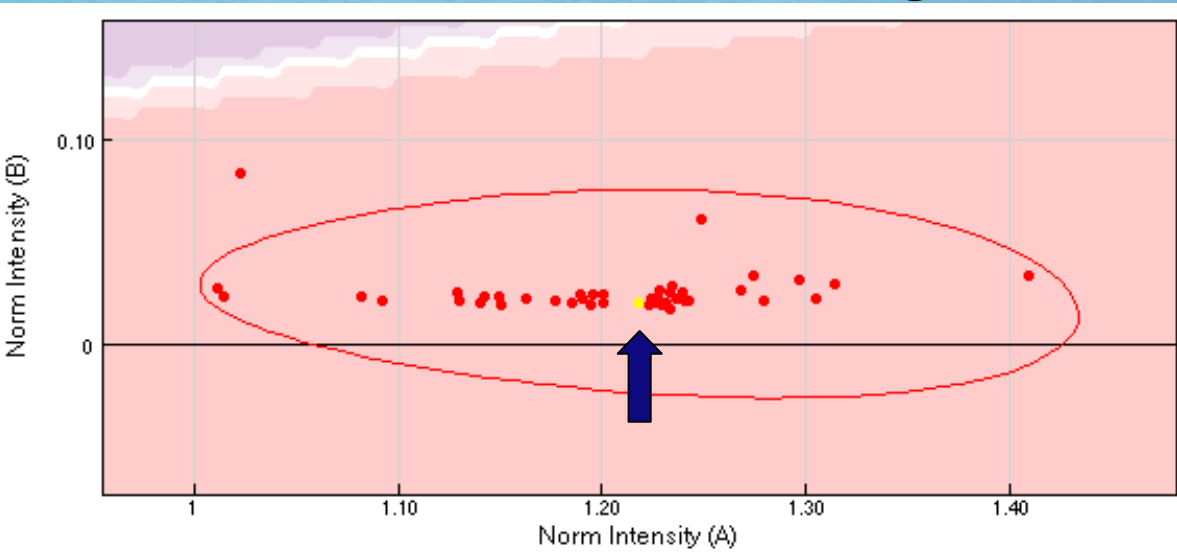
SNP 1: C1 500ng



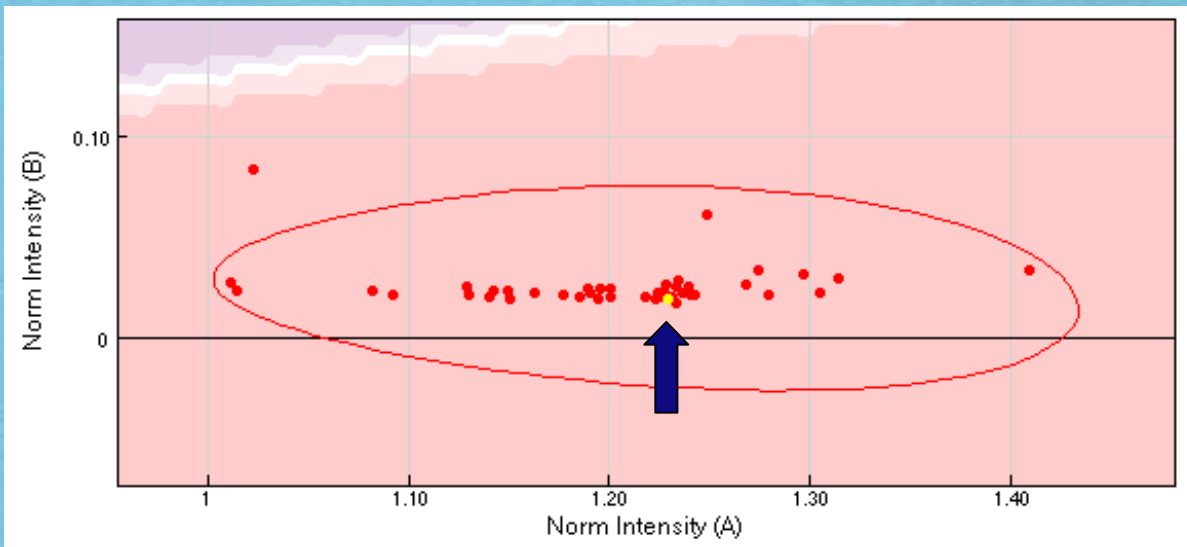
SNP 1: C1 500ng



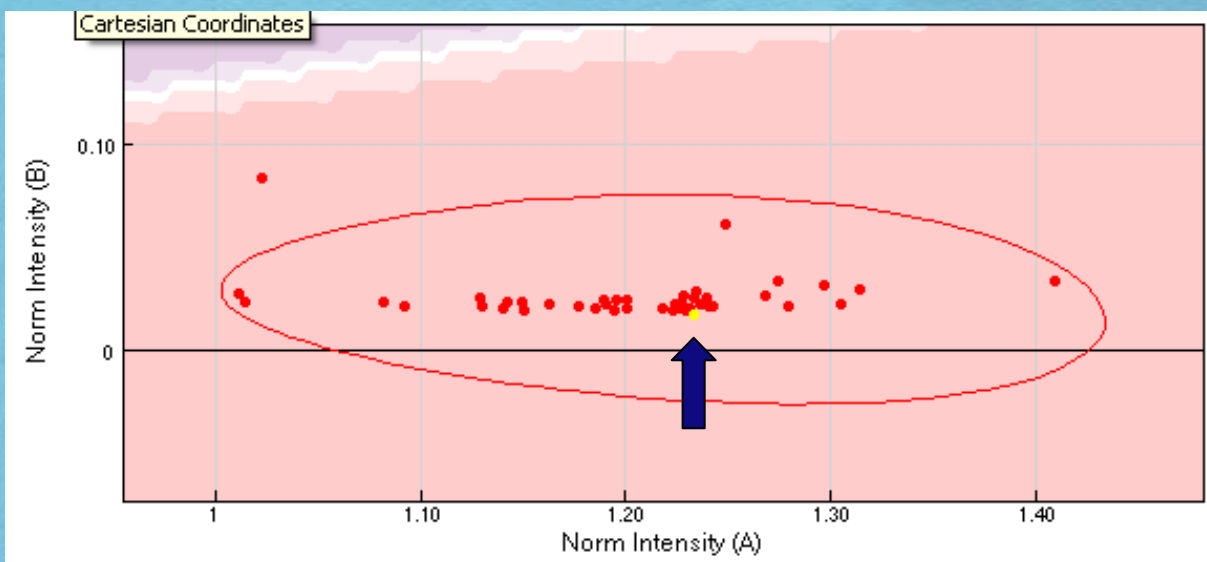
SNP 1: C1 250ng



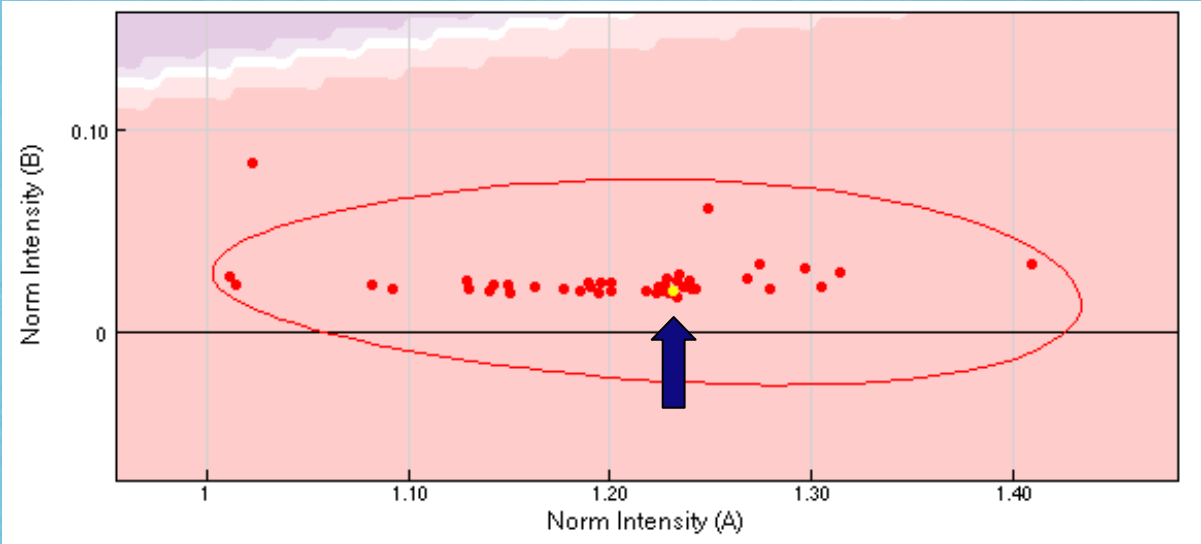
SNP 1: C1 125ng



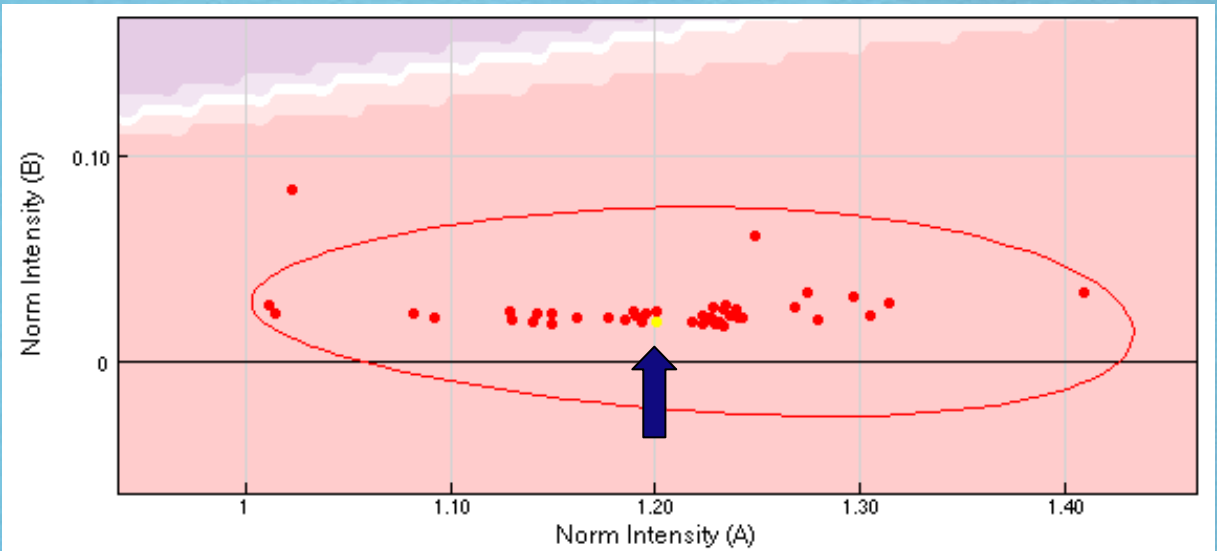
SNP 1: C1 62.5ng



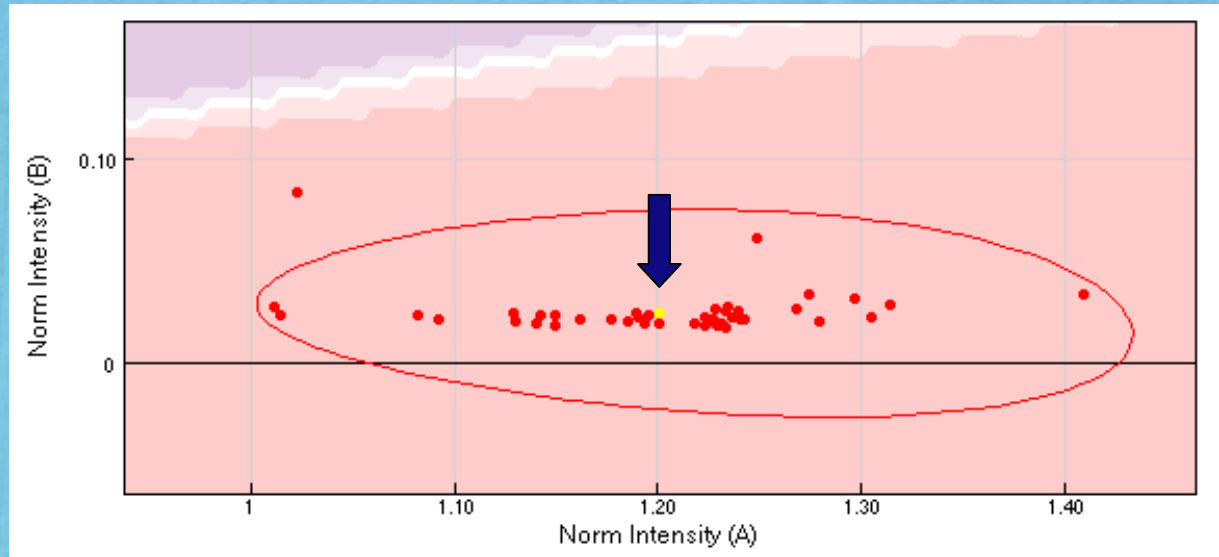
SNP 1: C1 31.25ng



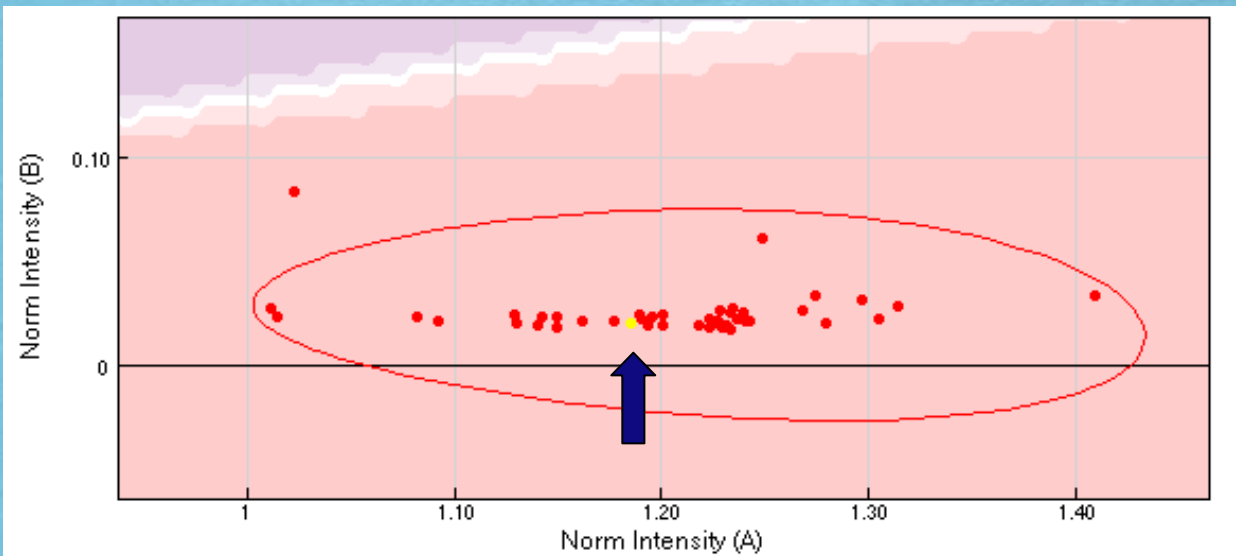
SNP 1: C1 15ng



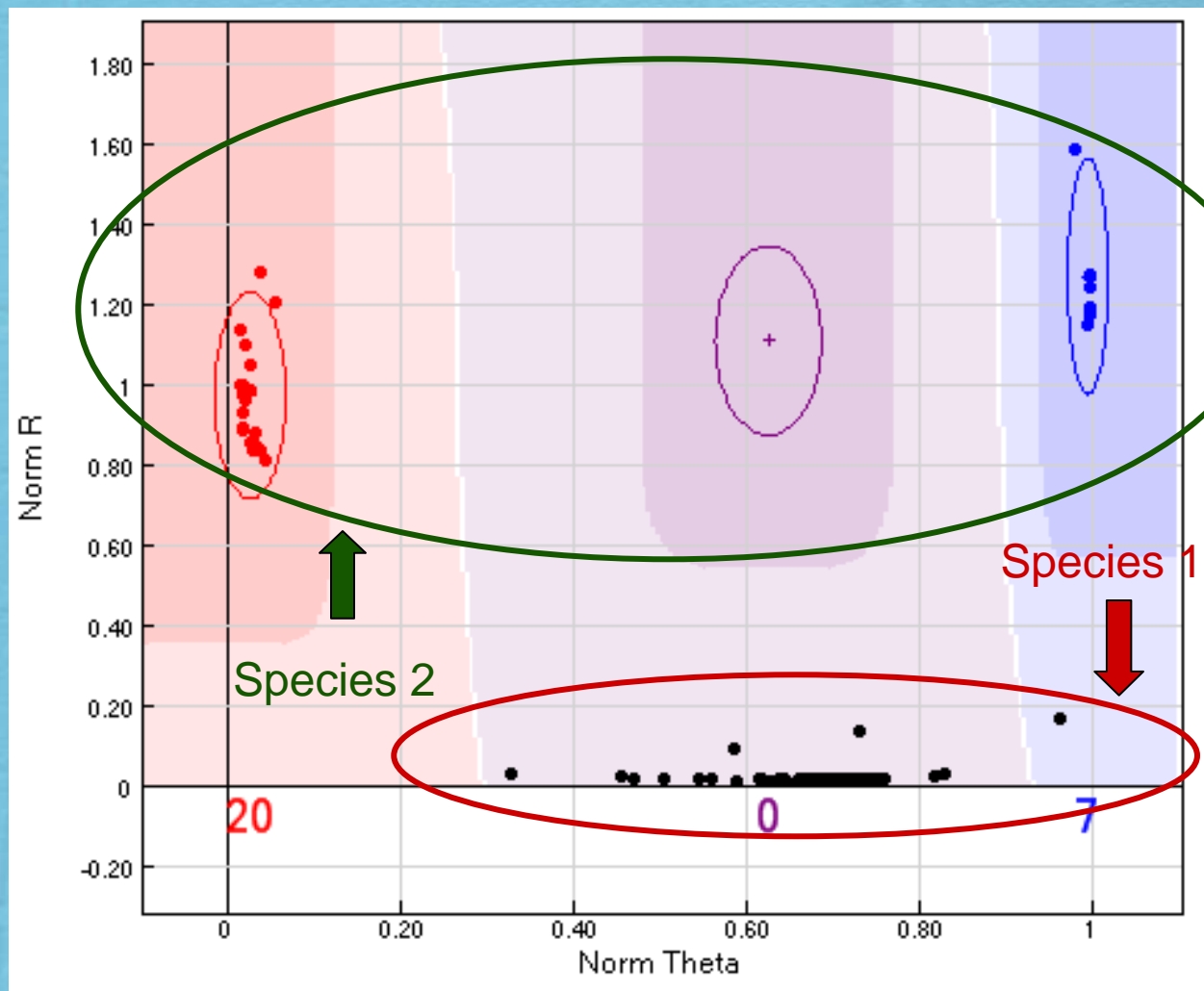
SNP 1: C1 7.5ng



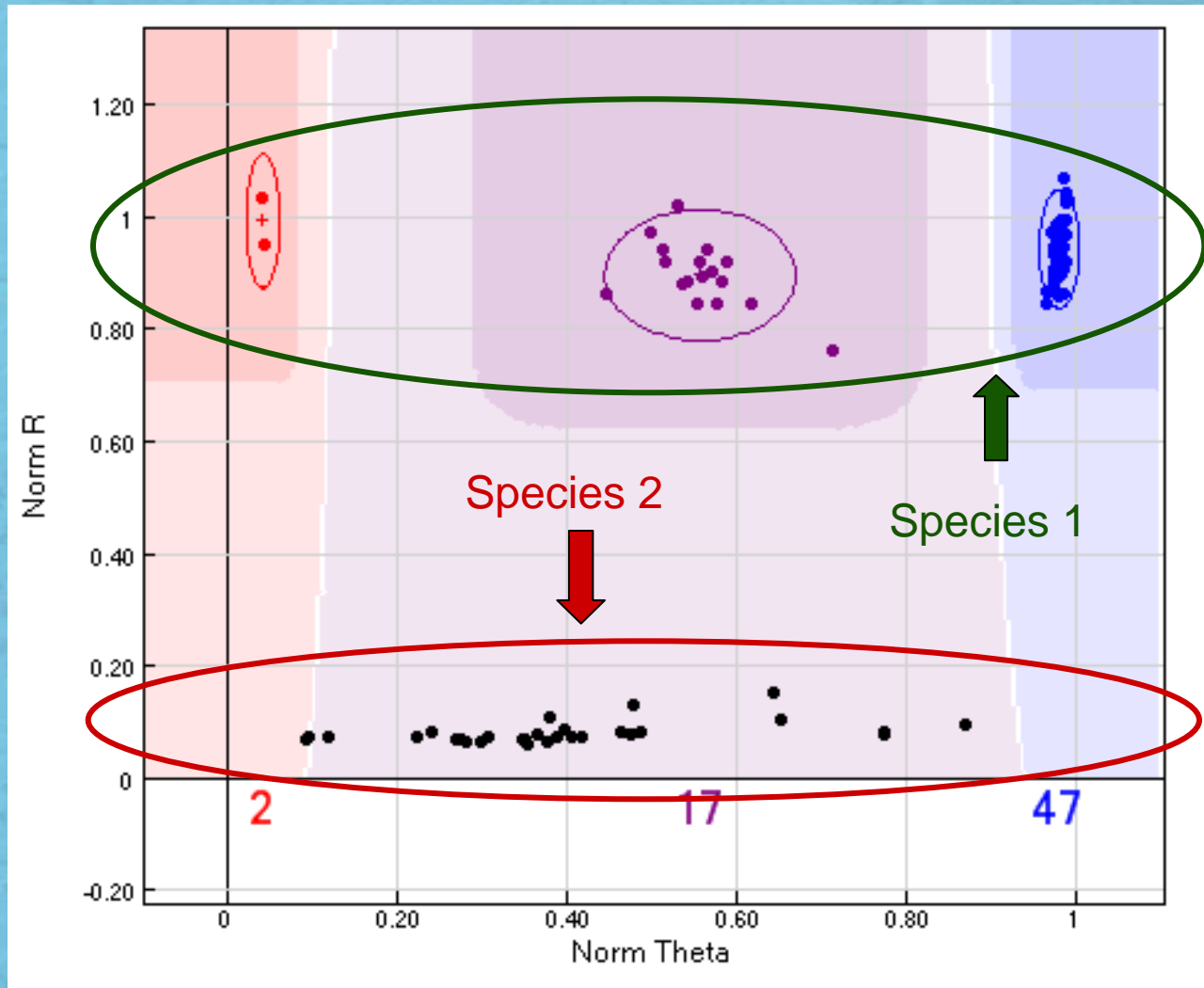
SNP 1: C1 3.75ng

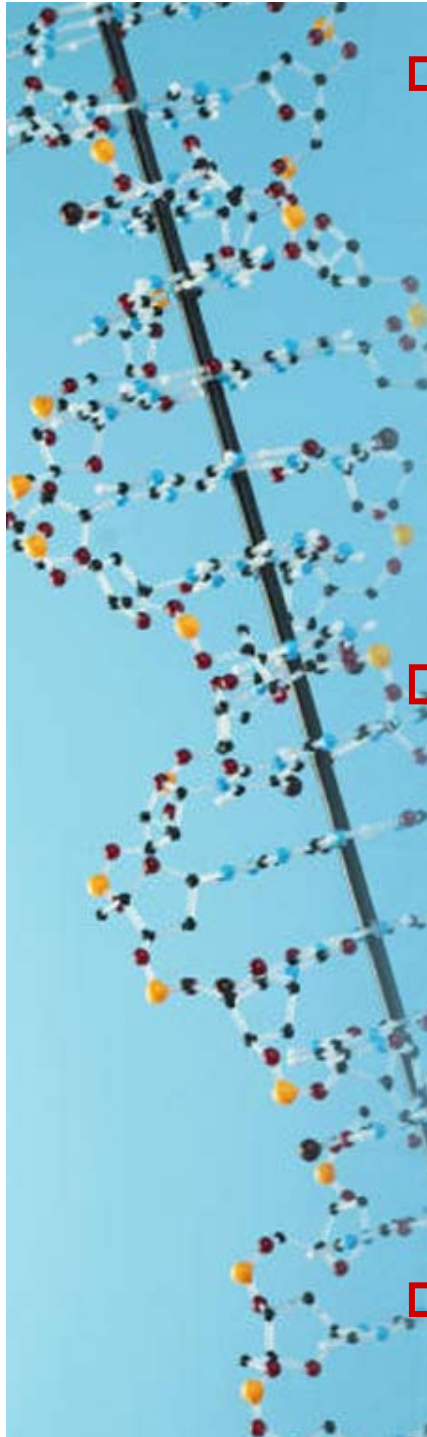


SNP Set 2: Species 2



SNP Set 1: Species 1





SNP	C1	S1	S1+ C1	H1	S1+H1	H2	S1+H2	H3	S1+H3	H4	S1+H4	H5	S1+H5
SNP1	G G	- -	G G	G G	A G	G G	A G	G G	- -	G G	A G	G G	A G
SNP2	A G	- -	A G	A G	A G	G G	- -	A G	A G	A G	- -	A G	- -
SNP3	A G	- -	A G	A G	A G	G G	G G	A G	A G	A G	A G	A G	A G
SNP4	G G	- -	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
SNP5	G G	- -	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
SNP6	G G	- -	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
SNP7	A A	- -	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
SNP8	A G	- -	A G	A A	A A	A A	A A	G G	G G	A A	A A	A G	A G
SNP22	A A	- -	A A	A G	A G	G G	G G	A G	A G	A G	A G	A A	A A
SNP23	A G	- -	A G	A A	A A	G G	G G	A G	A G	A G	A G	A G	A G
SNP24	G G	- -	G G	G G	G G	A A	A A	A G	A G	A G	- -	A A	A A
SNP25	A C	- -	A C	C C	C C	A A	A A	A A	A A	A C	A C	A A	A A
SNP26	C G	- -	C G	C C	C C	C G	C G	C G	C G	C G	C G	G G	G G
SNP27	G G	- -	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
SNP28	A A	- -	A A	A A	A A	C C	C C	A C	A C	A C	A C	C C	C C
SNP29	A G	- -	A G	A G	A G	A G	A G	A G	A G	A G	A G	A G	A G
SNP30	G G	- -	G G	G G	A G	G G	- -	A G	A G	G G	A G	G G	A G
SNP31	A A	- -	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
SNP32	A A	- -	A A	A G	A G	G G	G G	A A	A A	A G	A G	A A	A A
SNP33	A A	- -	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
SNP34	A A	- -	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
SNP35	A A	- -	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
SNP36	G G	- -	G G	A G	A G	G G	G G	G G	G G	A G	A G	A G	A G
SNP37	A A	- -	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A	A A
SNP38	G G	- -	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
SNP39	G G	- -	G G	G G	G G	G G	G G	G G	G G	A G	- -	G G	G G
SNP40	G G	- -	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G	G G
SNP41	C C	- -	C C	C C	C C	C G	C G	C C	C C	C C	C C	C C	C C
SNP48	G G	- -	G G	G G	G G	G G	- -	G G	G G	G G	G G	G G	G G
SNP49	G G	- -	G G	A A	A A	A A	A A	A G	A G	A A	A A	A A	A A
SNP50	C G	- -	C G	C C	C C	C G	C G	C C	C C	C G	C G	G G	G G
SNP52	C C	- -	C C	C C	C C	A A	A A	A C	A C	A C	A C	A A	A A
SNP53	A A	- -	A A	G G	G G	A A	A G	A A	A G	A A	A G	G G	G G
SNP54	G G	- -	G G	G G	- -	A A	A A	A G	A G	A G	A G	A A	A A
SNP55	A G	- -	A G	G G	G G	A A	A A	A A	A A	A G	A G	A A	A A
SNP56	A A	- -	A A	A G	A G	A A	A A	A G	A G	A A	A A	A A	A A



Conclusion

- Robust Assay
- Flexibility
- High Accuracy and Reproducibility
- High Call Rates
- Easy to integrate into LIMS