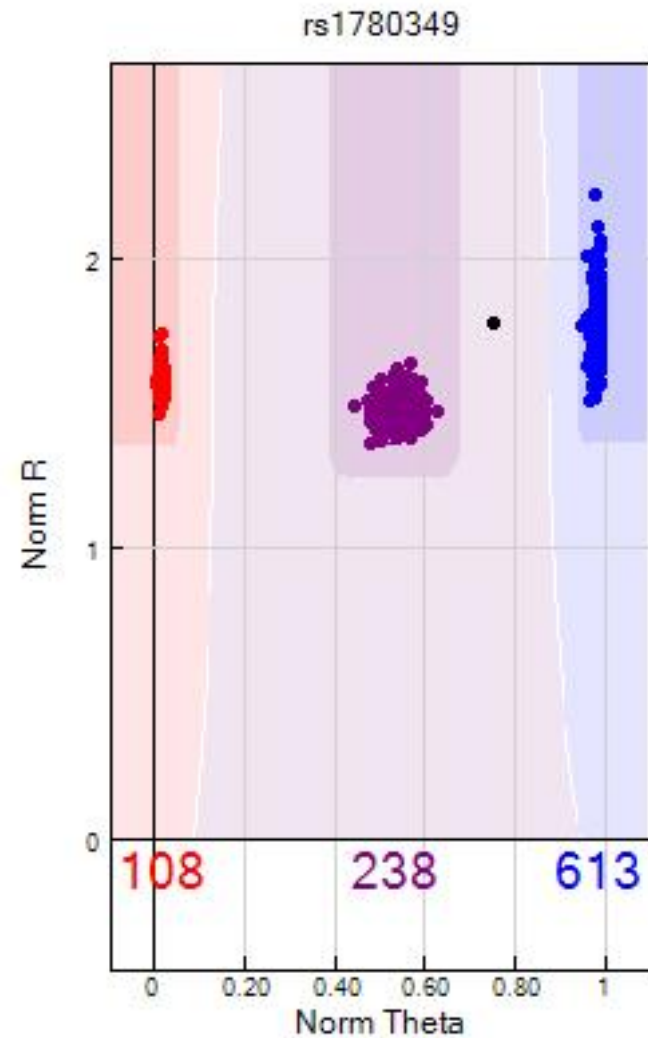
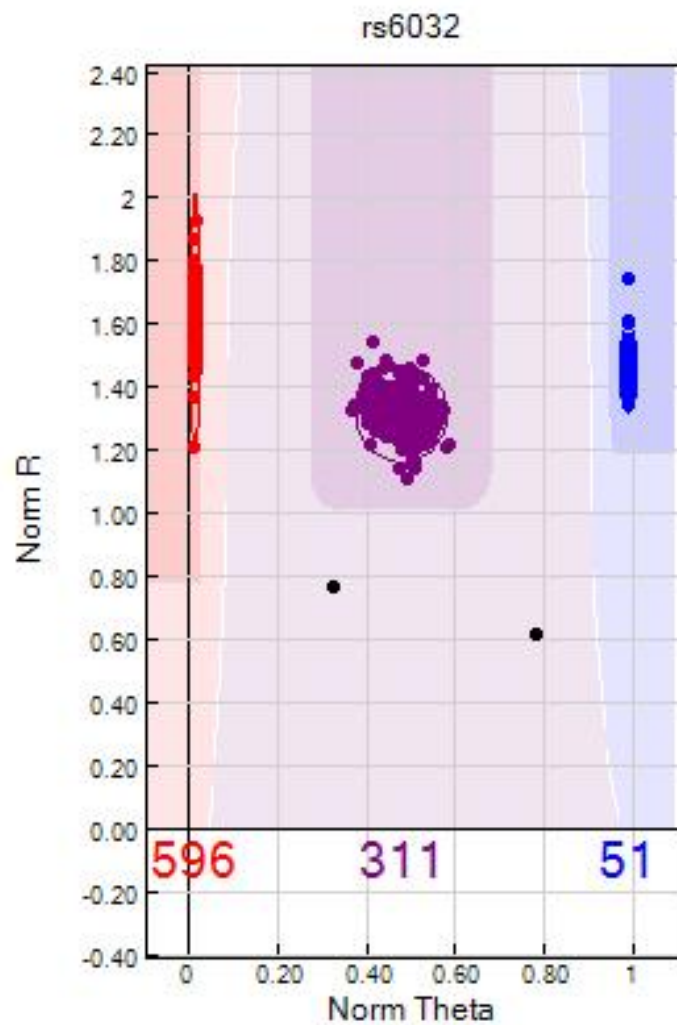
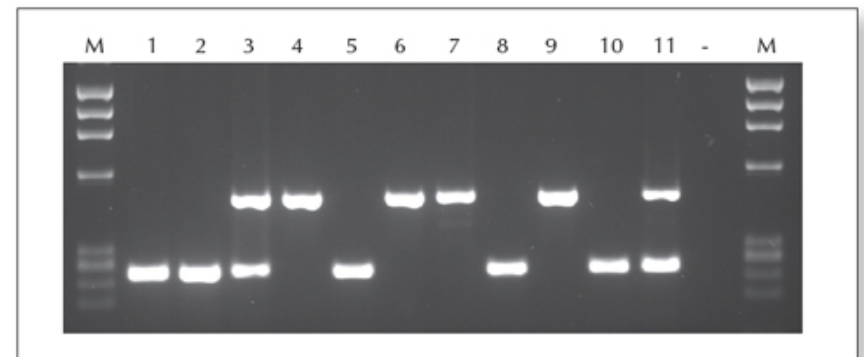


Genotype Clustering

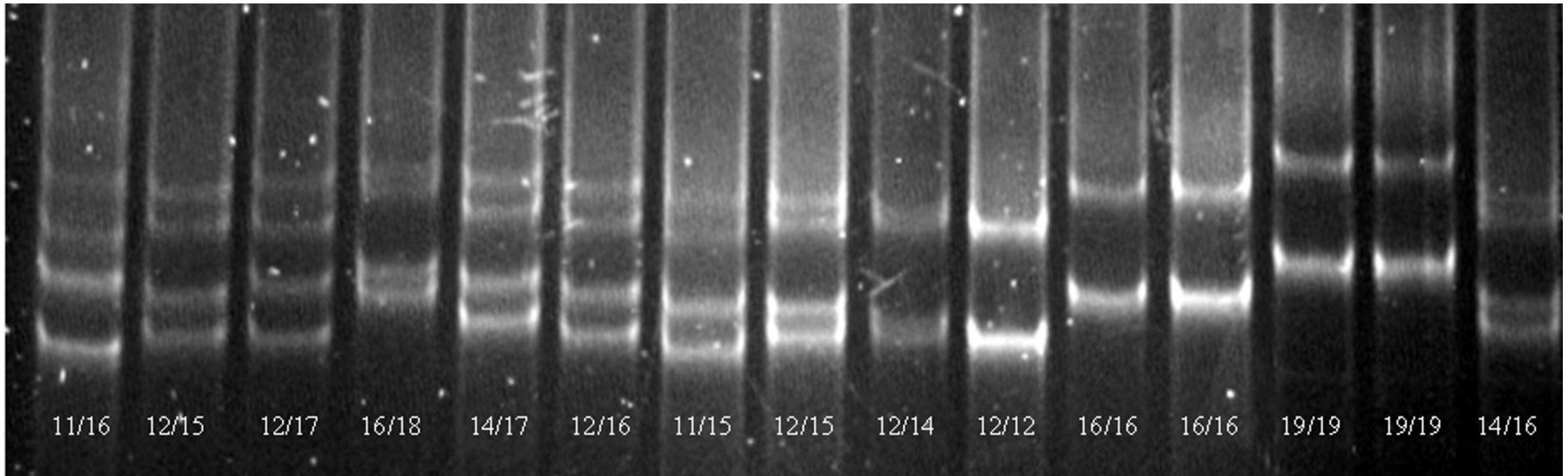


Sources of Genotype Calling Errors

- Incomplete enzyme digest
- Poor quality gel
- Poor data clustering
- Multiple clusters
- Missing alleles due to deletions



Poor Quality Gel



Poor Quality Gel

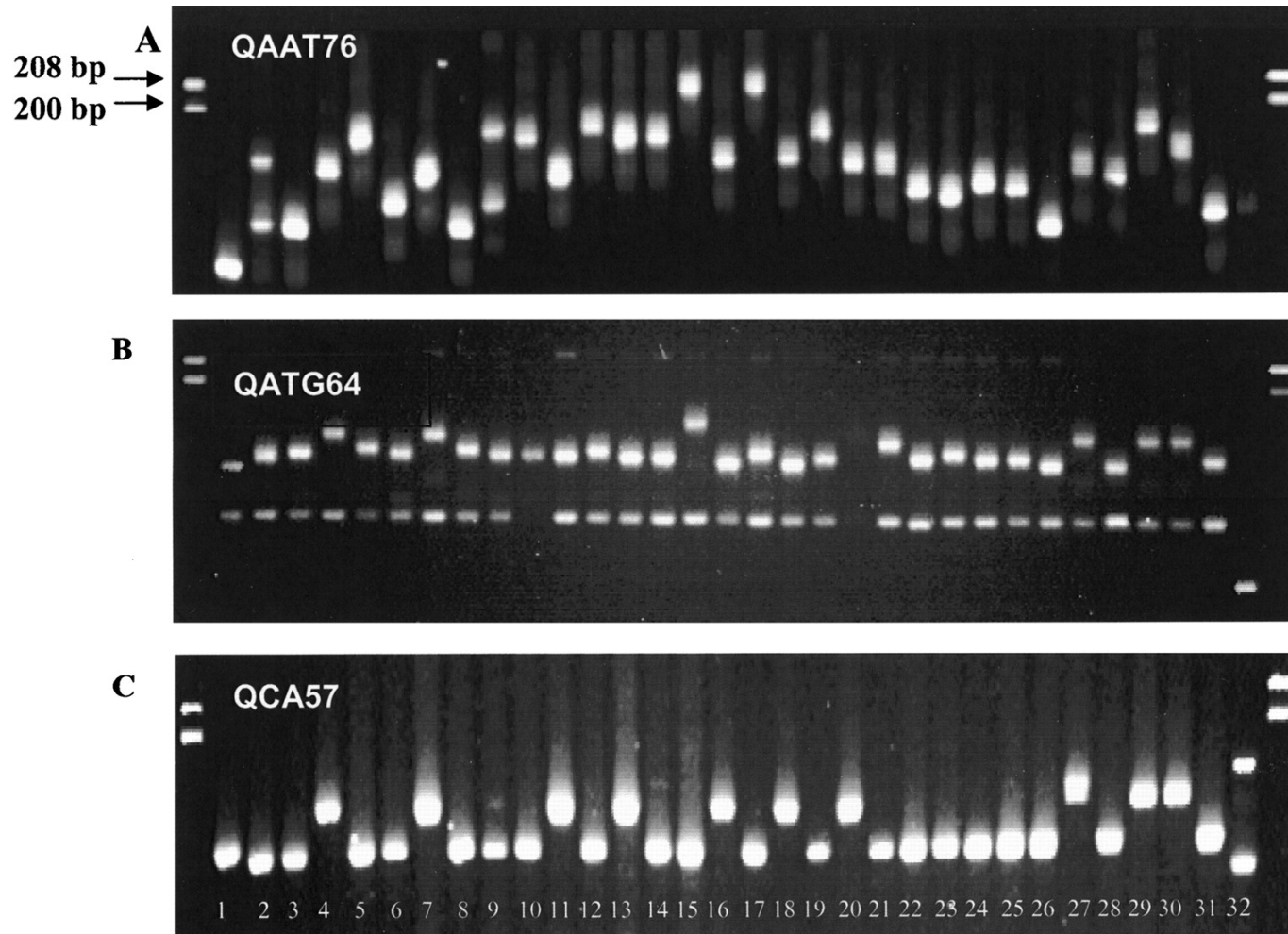


Figure 1. Compression of SNP Clusters

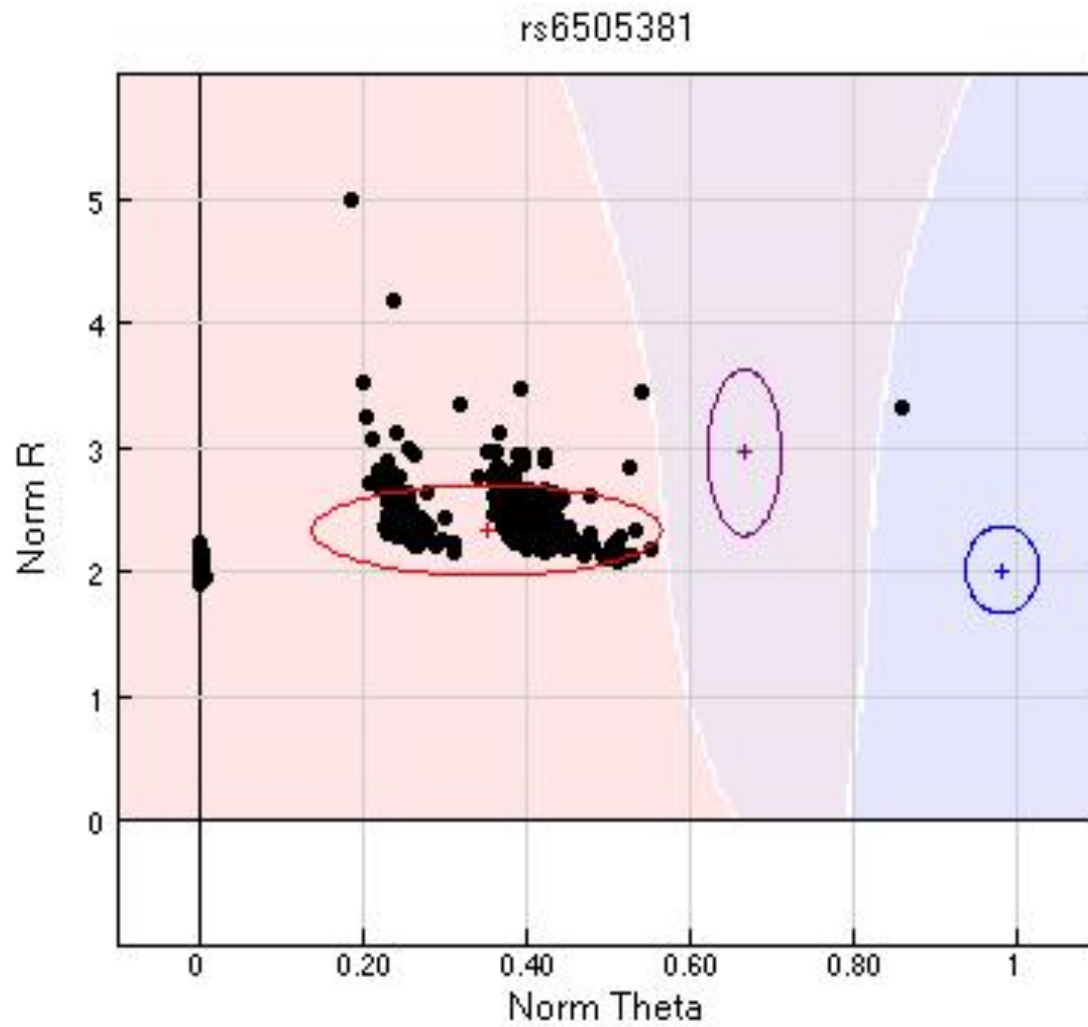


Figure 2. Low Intensity SNP

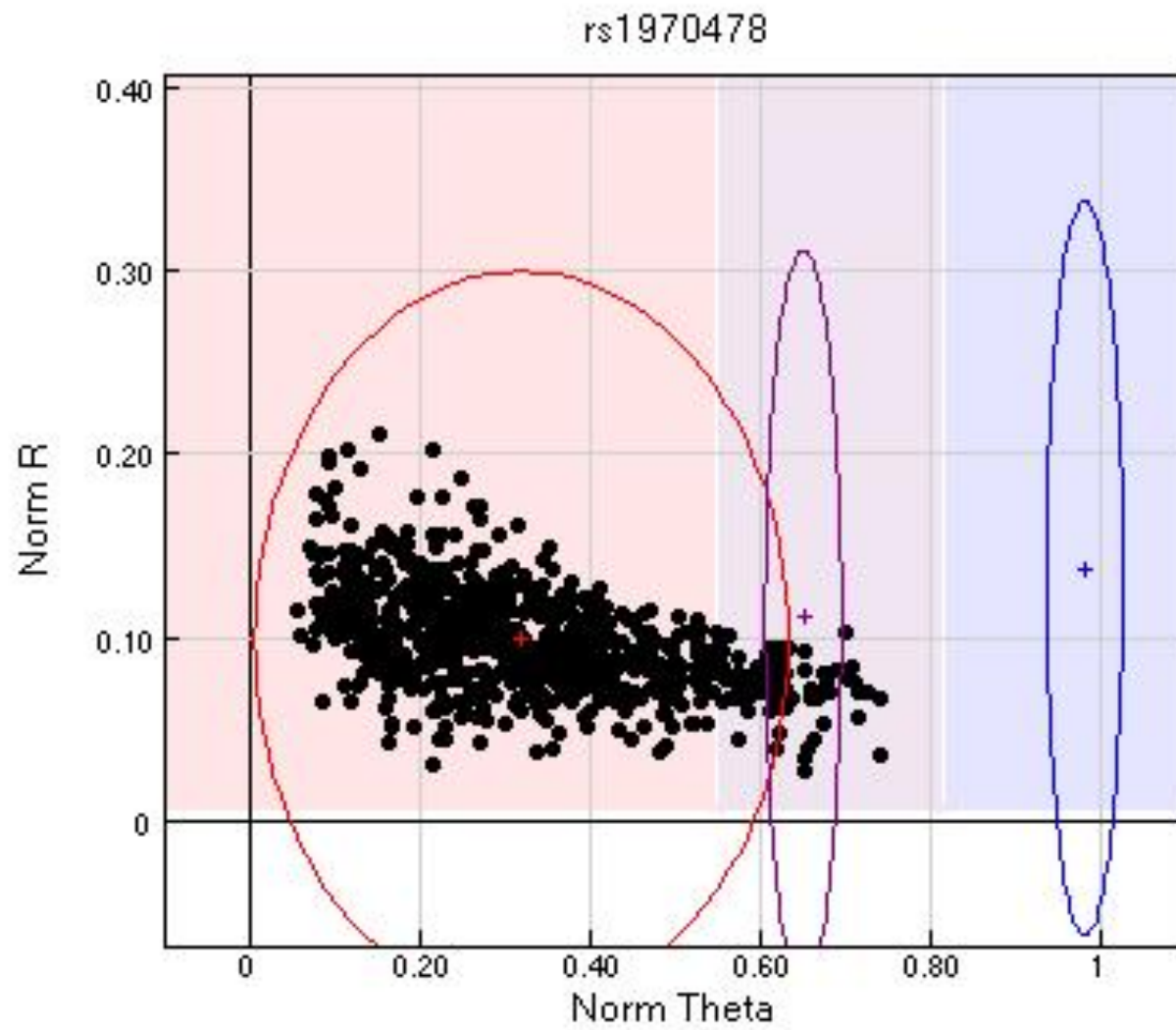


Figure 3. Multi Clusters

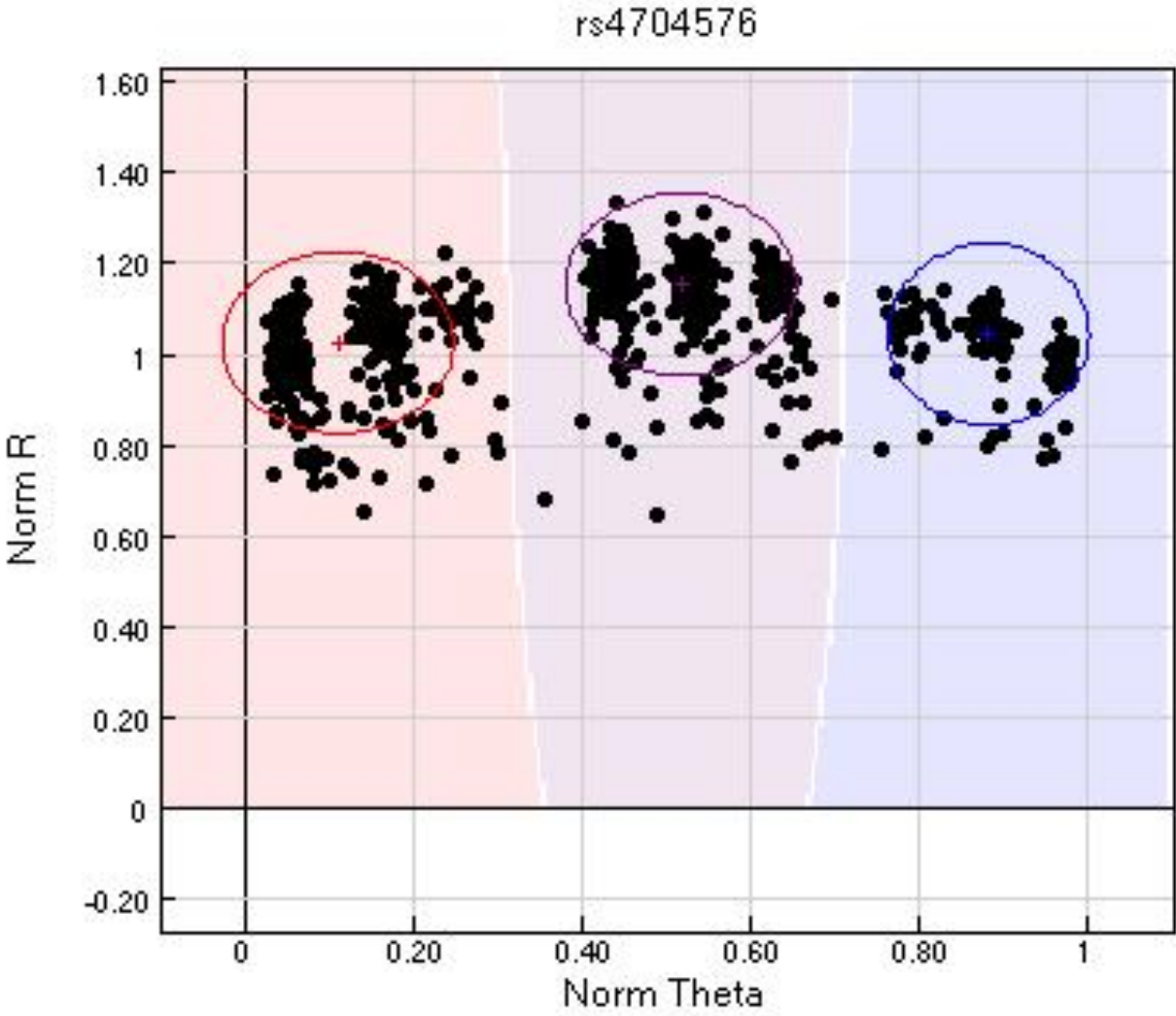
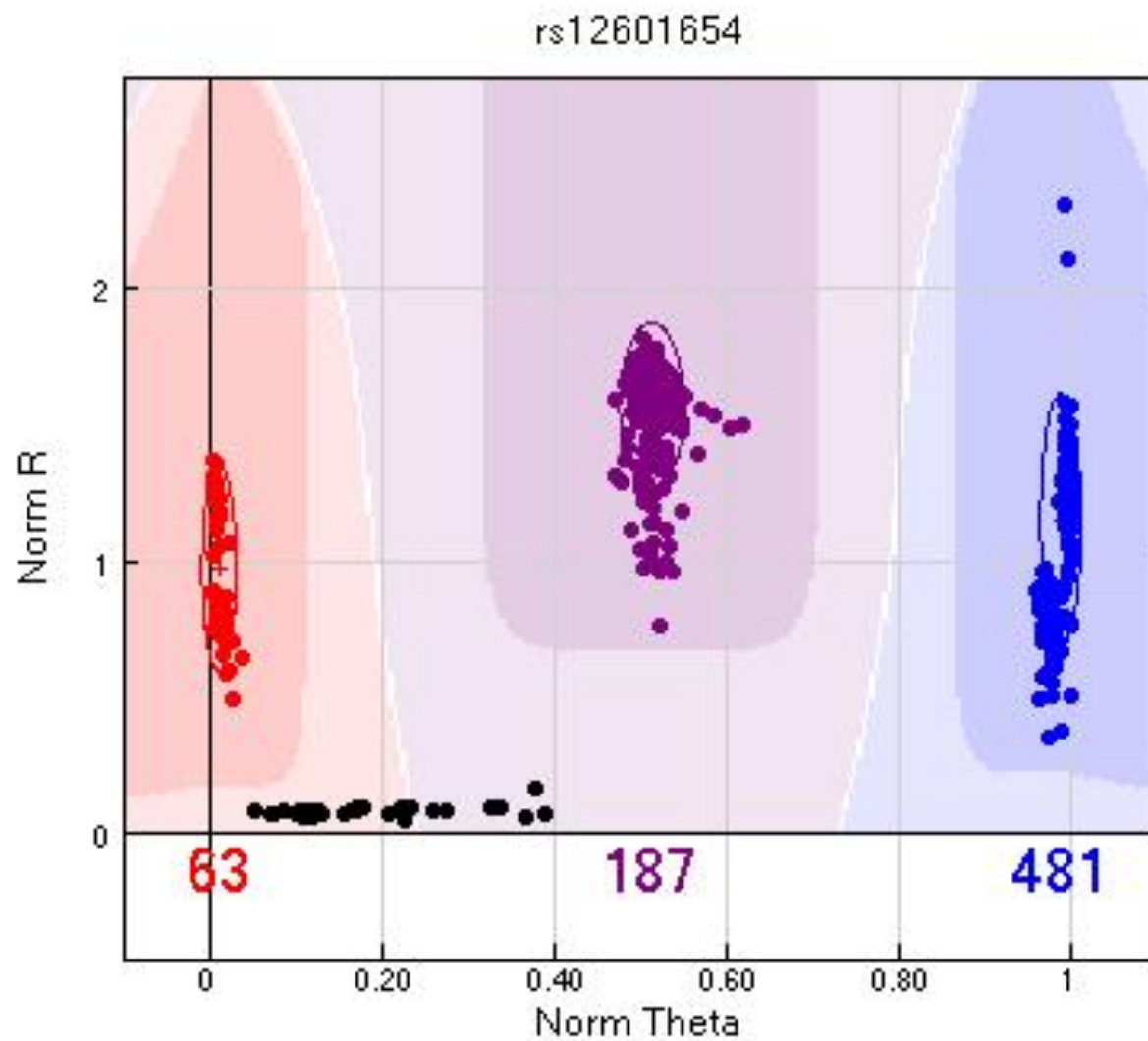


Figure 4. Potential Deletion

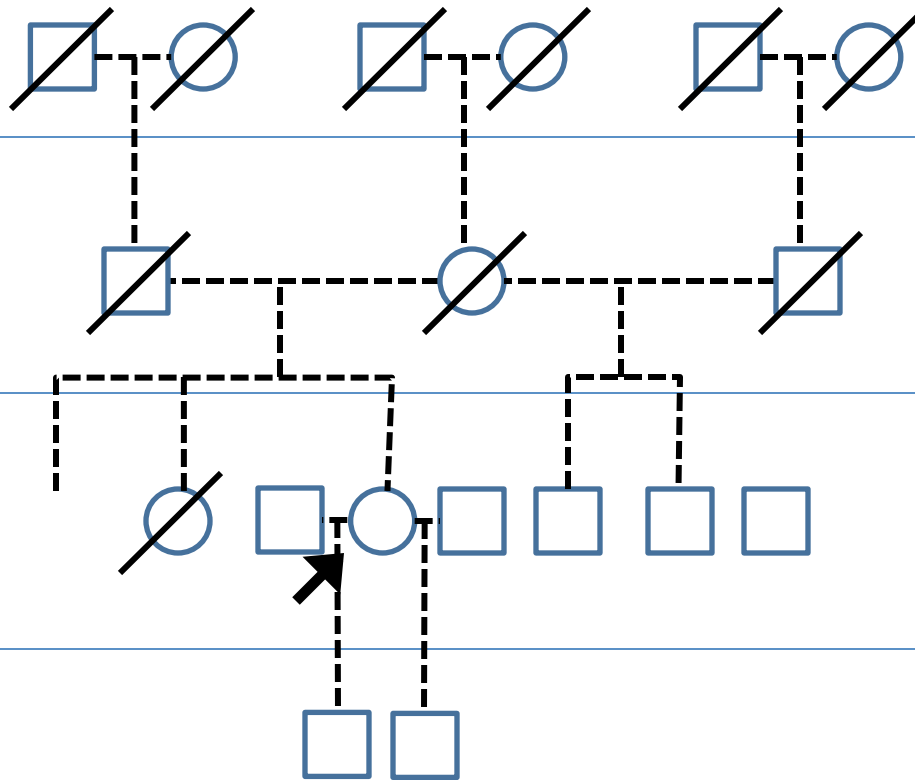


Draw Your Pedigree

Begin with the Proband – You!

Plan ahead – how many generations?

How complicated is the family?

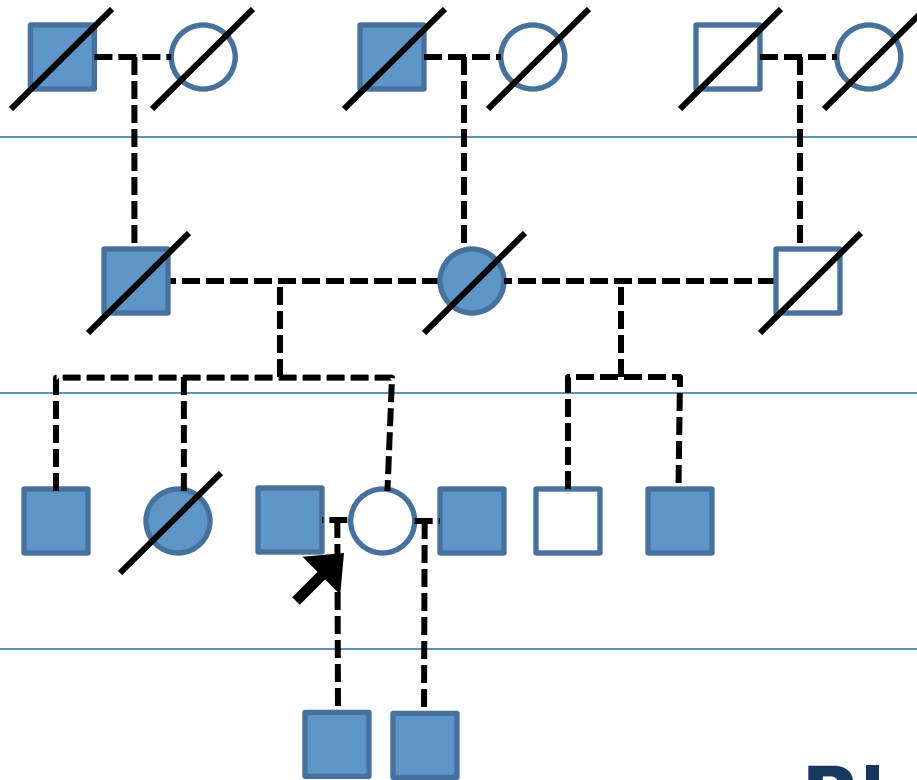


Draw Your Pedigree

Begin with the Proband – You!

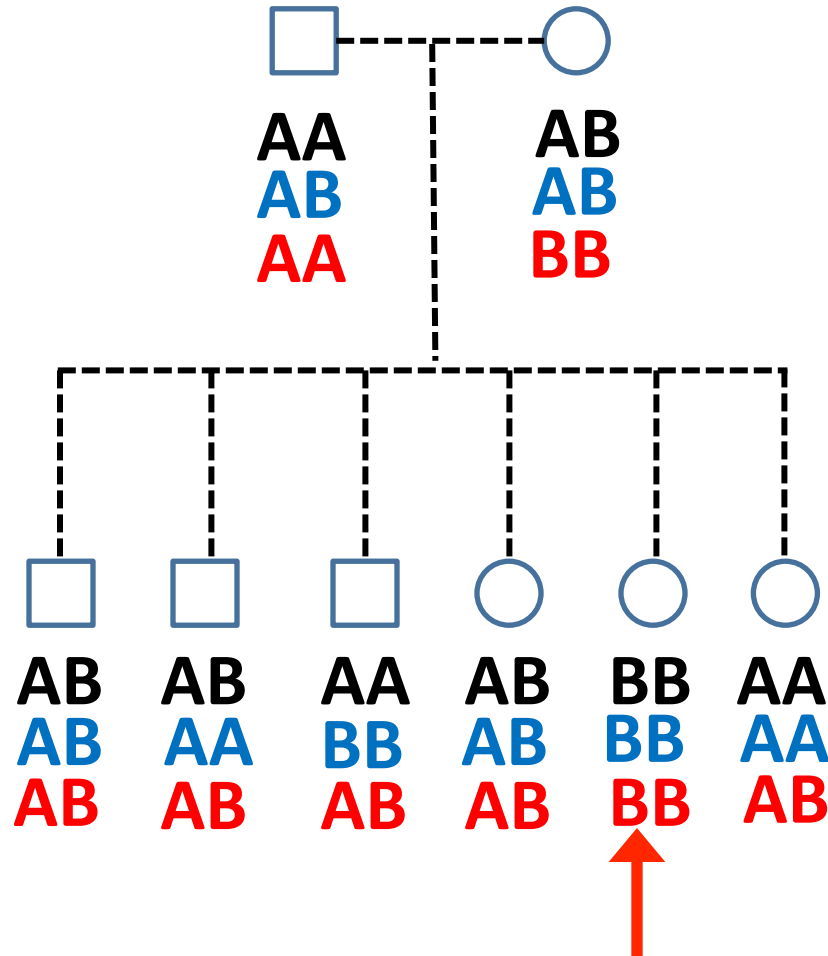
Plan ahead – how many generations?

How complicated is the family?



Blue Eyes

What can we tell about this family from genotype?



Who's The Daddy?
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Baltimore RH Lab
410-225-9595



Paternity Questions?
Answers available
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IDENTIGENE
Since 1991

DNA PATERNITY TEST

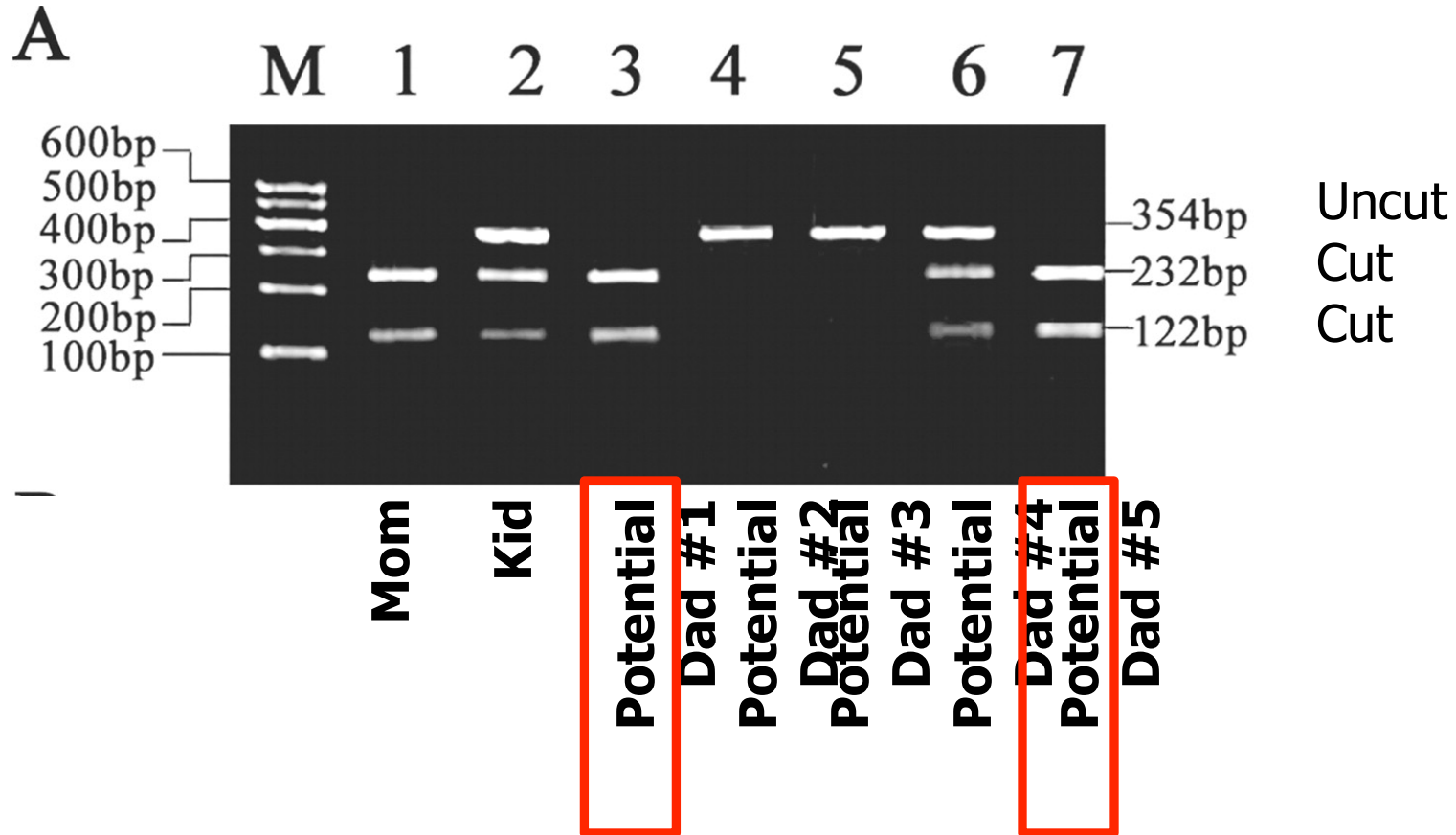
COLLECTION KIT
For Alleged Father, Mother and Child

Results Typically Reported in 2 Days



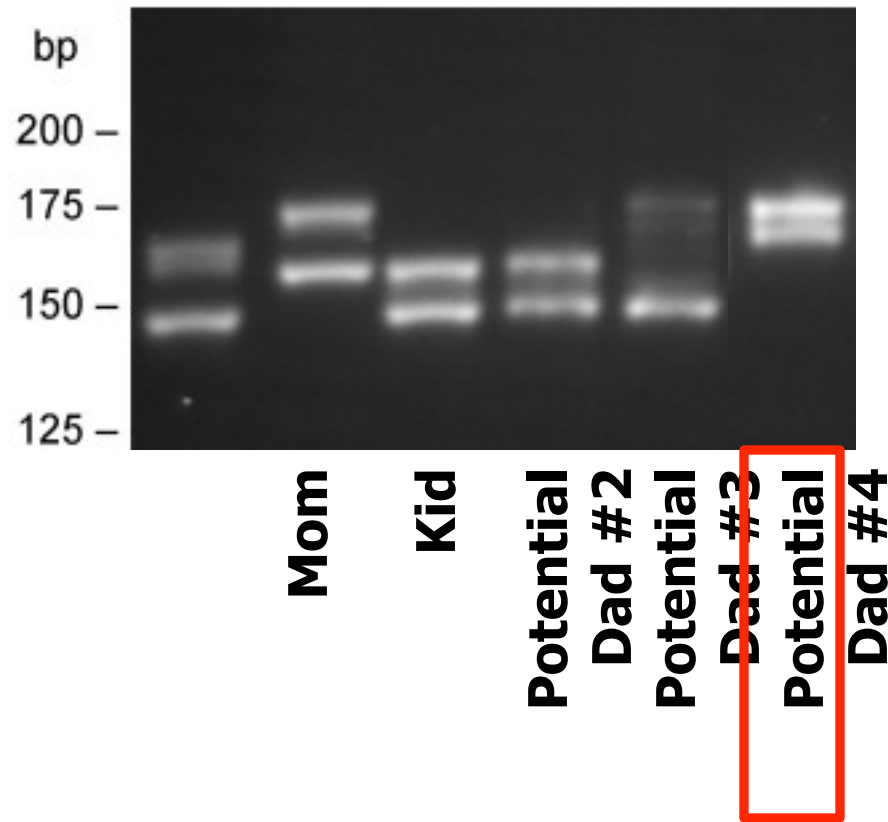
“In a few days, you’ll know whether you’re either stuck with that kid for a few more decades or will once again taste the sweet nectar of freedom.”

Restriction Fragment Length Polymorphism



Can you rule out any of these potential dads?

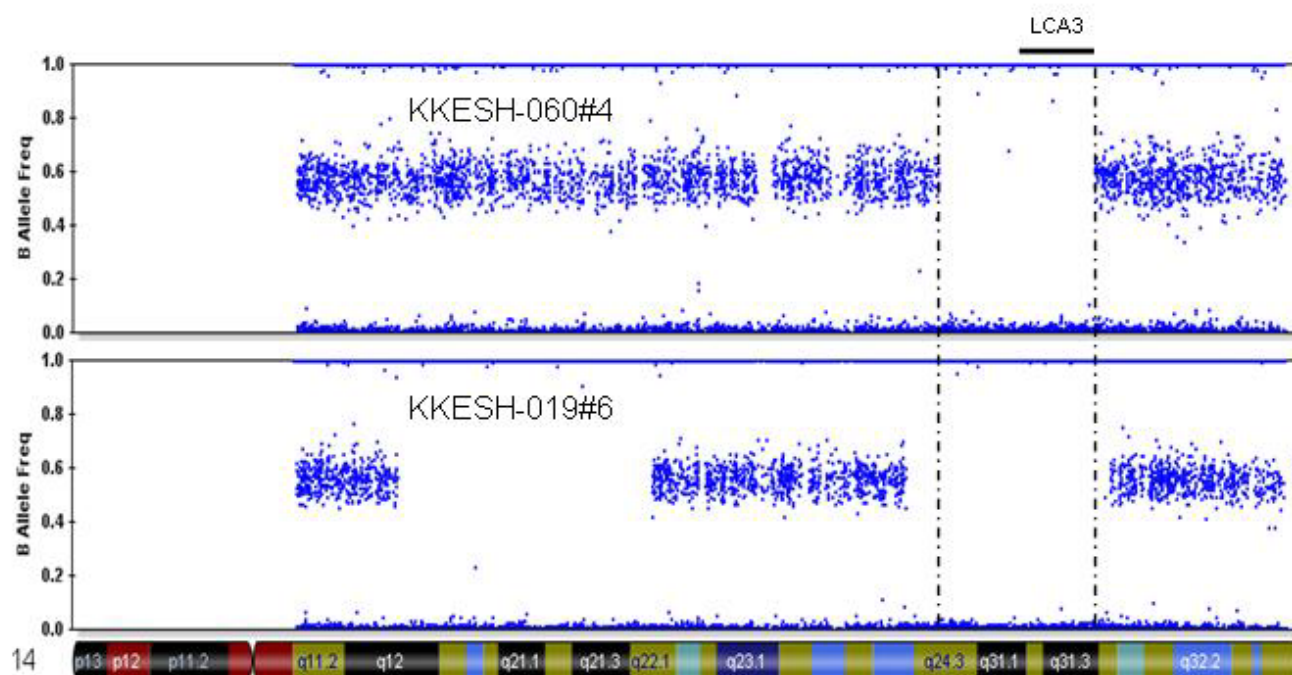
Microsatellite Markers



Can you rule out any of these potential dads?

Mutations in *SPATA7* Cause Leber Congenital Amaurosis and Juvenile Retinitis Pigmentosa

Hui Wang,^{1,7} Anneke I. den Hollander,^{10,11} Yalda Moayed, Yalda Moayed,³ Abuduaini Abulimiti,^{1,7} Yumei Li,^{1,7} Rob W.J. Collin,¹⁰ Carel B. Hoyng,¹¹ Irma Lopez,¹² Molly Bray,⁸ Richard Alan Lewis,^{1,2,9} James R. Lupski,^{1,5,9} Graeme Mardon,^{1,2,3,4,6} Robert K. Koenekoop,¹² and Rui Chen^{1,6,7,*}



Conclusions

- DNA sequence variation contributes to diversity of species.
- Types of DNA sequence variation include SNPs, insertion/deletions, microsatellites, and copy number variation.
- Many options for genotyping take advantage of natural DNA machinery.
- Mapping DNA sequence variation to human traits and disease is challenging but new technologies are advancing genomic science.