

# Genetics in PPMI

Andrew Singleton



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Play a Part in Parkinson's Research

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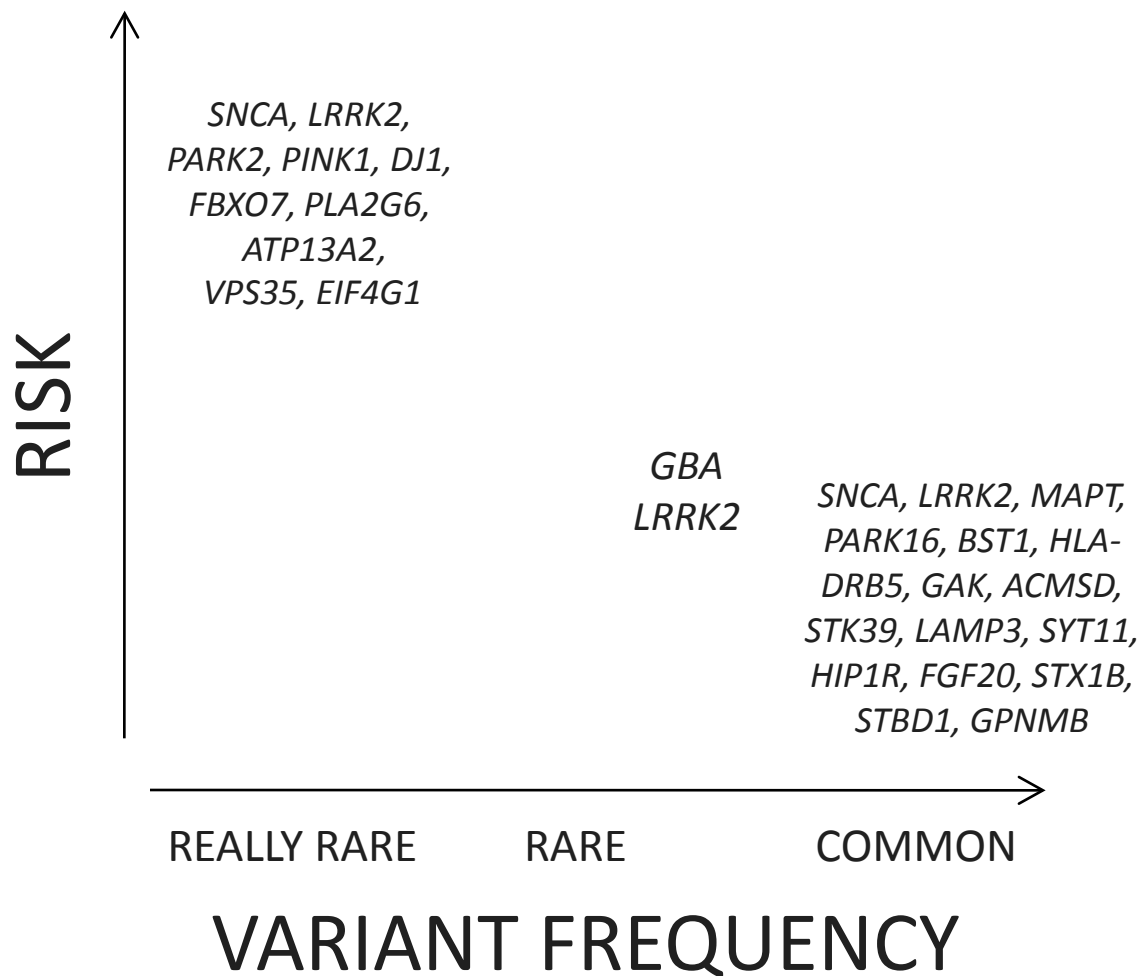


# Genetics in PPMI

- Probably not used as a discovery cohort for genetics by itself
- Most useful as a covariate in the biomarker studies
- Incredibly valuable with other studies on disease subtype (progression etc)
- Immediate availability of data a boon



# Genetic Architecture of PD

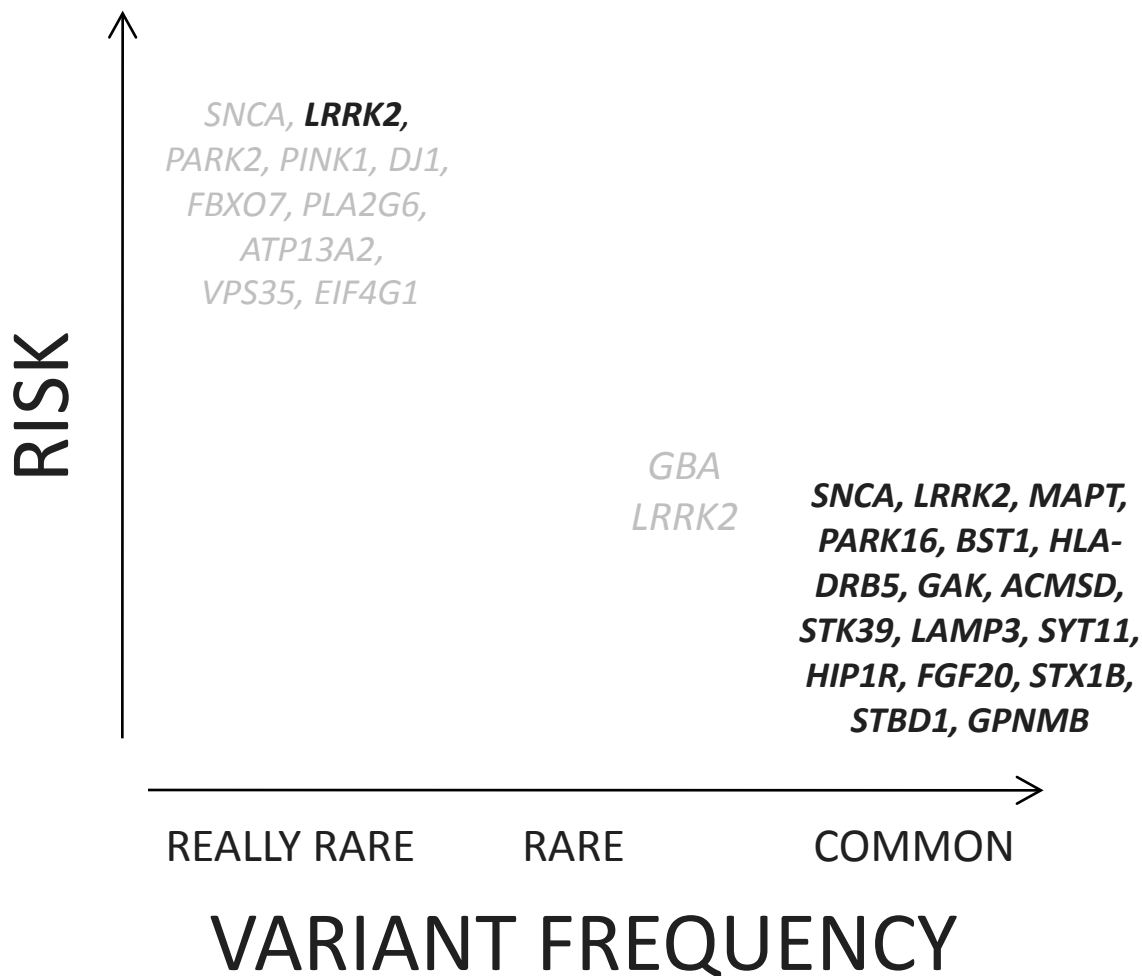


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# ImmunoChip



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# Data so far

- First 100 samples received in early 2012
- Initial DNA QC all passed
- 96 samples run on the ImmunoChip
- Post genotype QC – 3 samples with mismatched gender (agreed with Coriell QC)
- Sample success rate – 100%
- Genotype success rate – 98.7%
- Data uploaded to LONI

**all proceeding as planned**



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# Next Steps

- Will continue with ImmunoChip through all samples
- Include individual genotyping (APOE, DAT vntr etc)
- Augment with exome genotyping (neuroX?)
- Likely some addition of sequence data (exome, resequencing, genome)
- Challenge in timing of technology and cohort collection

