Useful Genetics Module 6 Overview—Personal genomics

Reference Abbreviations
OG: Open Genetics
OSB: Open Stax Biology
IGAG: Introduction to Genetic Analysis, 10th Ed. (Griffiths et al.)

IGAG: General reading on gene identification, genomes and genomics: 10.3-10.5, Ch 14

6A – DNA fingerprinting
Outline
- Why DNA makes good evidence
- 'VNTR' loci have many alleles
- New combinations in each new person
Learning objectives
- Explain why DNA provides valuable evidence
Reading resources
OG: 10.4
OSB: 17.2 “Genetic maps”
IGAG: none

6B – VNTR markers in DNA analysis
Outline
- VNTR mutations arise often, when DNA polymerase slips (but not too often)
- The CODIS set of VNTR loci was developed for forensic work
Learning objectives
- Illustrate alleles of a VNTR locus
- Explain the importance of the balance between VNTR variation and stability
Reading resources
(See readings for Lecture 6A)

6C – Analyzing a single gene or small gene panel
Outline
- What can be learned
- Availability
- Limitations and concerns
Learning objectives
- Distinguish between what can be learned from single-gene testing and what cannot
Reading resources
OG: General reading on gene identification, genomes and genomics: 10.3-10.5
OSB: General reading on gene identification, genomes and genomics: 17.2-17.6

6D – Examples of single gene analysis
Outline
- Disease risk
- Pre-implantation screening
- Tumor typing
- Cancer risk
- What about BRCA alleles?
Learning objectives
- Describe situations where single-gene testing is appropriate
Reading resources
OG: none
OSB: 17.6 “Predicting disease risk at the individual level”
IGAG: none

6E – SNP-typing the genome (the science) Part 1: HapMap
Outline
- Reminder: SNPs
- How HapMap came about
- A guided tour of HapMap
Learning objectives
- Use HapMap to examine human SNP variation
- Say 'Thank you' to the taxpayers who paid for HapMap
Reading resources
OG: 10.1-10.4
OSB: 17.2 “Genetic maps”
IGAG: 4.3, 18.1

6F – SNP-typing the genome (the science) Part 2: Predicting personal phenotypes
Outline
- GWAS data connects SNP genotype to phenotype (odds ratios)
- SNP typing predicts individual phenotype probabilities
Learning objectives
- Distinguish between causal and non-causal SNPs
- Interpret information about relative risk
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Reading resources
OG: none
OSB: 17.6
IGAG: 19.6

6G – How SNP-typing works
Outline
- How SNP-typing chips work

Learning objectives
- You don't really need to know how SNP-typing chips work
- But you should be amazed that this technology is so cheap

Reading resources
None

6H – Personal SNP-typing: information about phenotype
Outline
- Who provides this?
- What do they offer?
- Features to look for (it's all about the interpretation)

Learning objectives
- Evaluate personal SNP-typing services as sources of health and other phenotypic information

Reading resources
None

6H* – Using raw SNP-typing data
Outline
- What raw data is
- Who would provide it to you
- Accessing public SNP data
- What you can do with it

Learning objectives
- Describe raw SNP data and ways it can be used

Reading resources
None

6I – Thinking about ancestry
Outline
- Many companies offer ancestry analysis
- Visualizing ancestors
- Visualizing many many relatives

- How many ancestors do we have?

Learning objectives
- Use diagrams to describe ancestry and relationships
- Explain why we don't have $2^n$ ancestors

Reading resources
None

6J – Ancestry analysis: mitochondrial and Y-chromosome DNAs
Outline
- Mitochondrial DNA tracks only the maternal lineage
- Y-chromosome DNA tracks only the paternal lineage
- Most ancestors are not detected

Learning objectives
- Distinguish between different types of ancestry analysis
- Distinguish between haplotype and haplogroup

Reading resources
OG: none
OSB: none
IGAG: 2.6 “Y-linked inheritance”, 3.5

6K – Exome and whole-genome sequencing
Outline
- Why would anyone do this?
- How you can, how I did
- What I learned

Learning objectives
- Explain what an 'exome' is
- Compare what can be learned from SNP typing and from exome or whole-genome sequencing

Reading resources
None

6L – Ethical and social issues
Outline
- What difference will this information make to me? To my family?
- Who will help me understand it?
- What if it's bad news? What if it's good?
- Who will I share this with? Who not?
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- Is this even legal?

Learning objectives
- Present reasons for and against obtaining particular genetic information

Reading resources
None

6M – Not-so-personal genomics

Outline
- Confirming identity and parentage of valuable animals (dogs, horses, cattle, sheep)
- Predicting performance of livestock
- Breed characterization (dogs...)
- Sex-typing (birds)

Learning objectives
- Think of ways you can use genomics to make lots of money!

Reading resources
none