

# **Forensic Genetics and Legal Medicine 2019-2020**

**4th May 2020**

**Beyond identification  
(intro and sex-typing)**

# Woody Allen

“Everything  
you always  
wanted to know

from forensic genetics

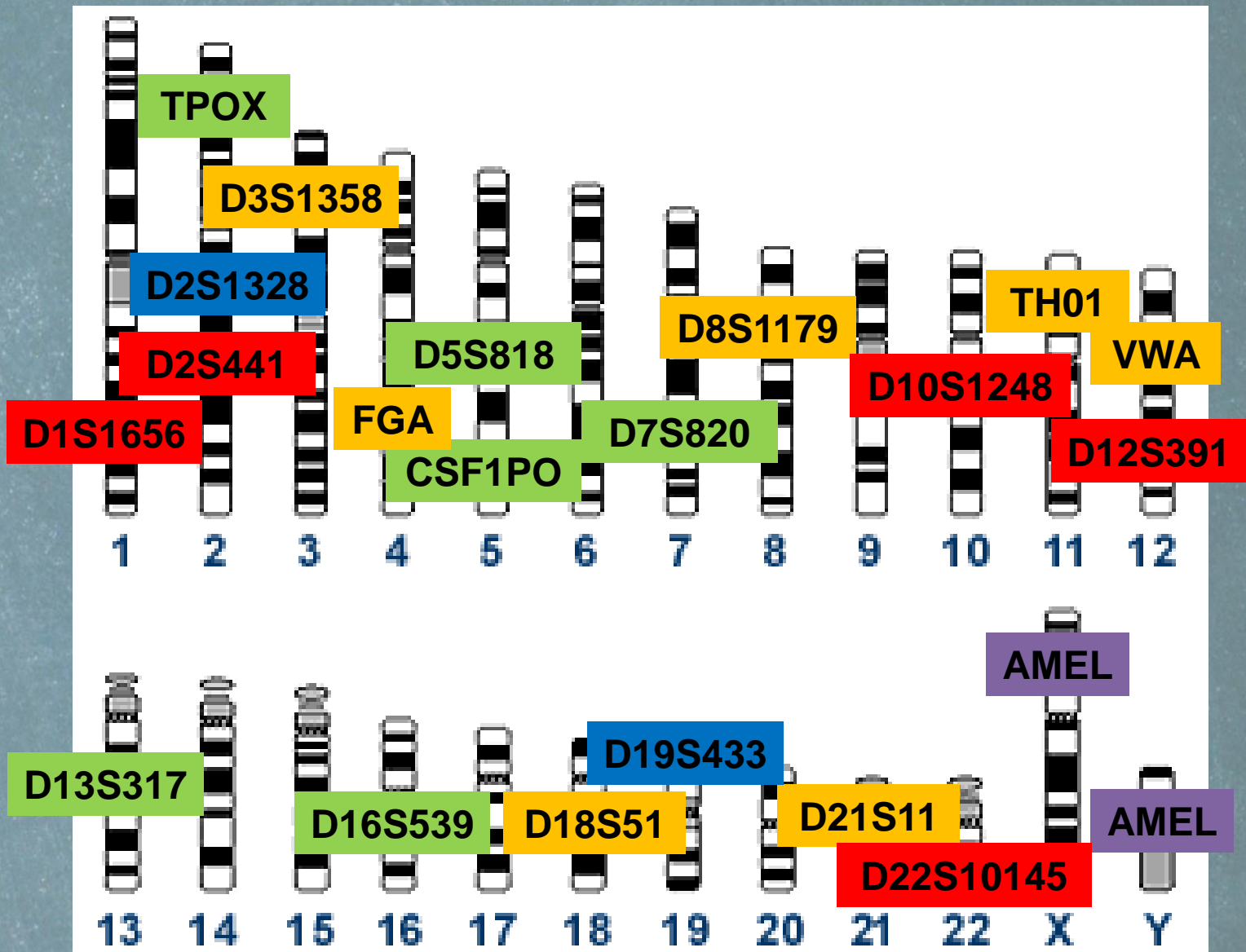


\* BUT WERE AFRAID  
TO ASK”



✓ Externally visible characters derived from DNA data that could help identifying the unknown donor of a stain («forensic phenotyping»)

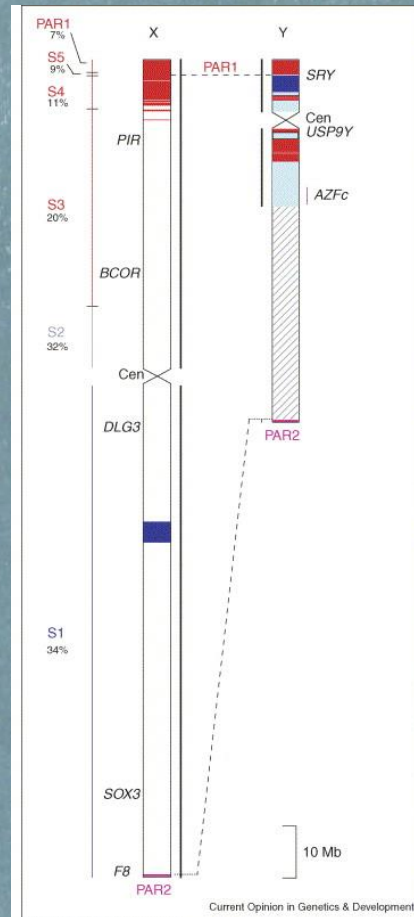
- sex
- biogeographical ancestry
- Pigmentation (eye, hair, skin color)
- age



# Sex

## X chromosome

- ~ 165 Mb
- ~ 1000 genes

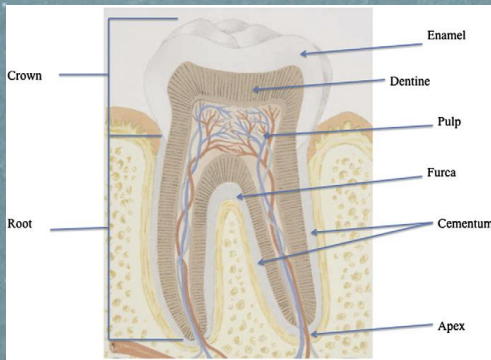


## Y chromosome

- ~ 65 Mb
- ~ 50 genes
- ~50% of genes in the male specific region (MSY)
- ~50% of genes in the pseudoautosomal region (PAR) homologous to X



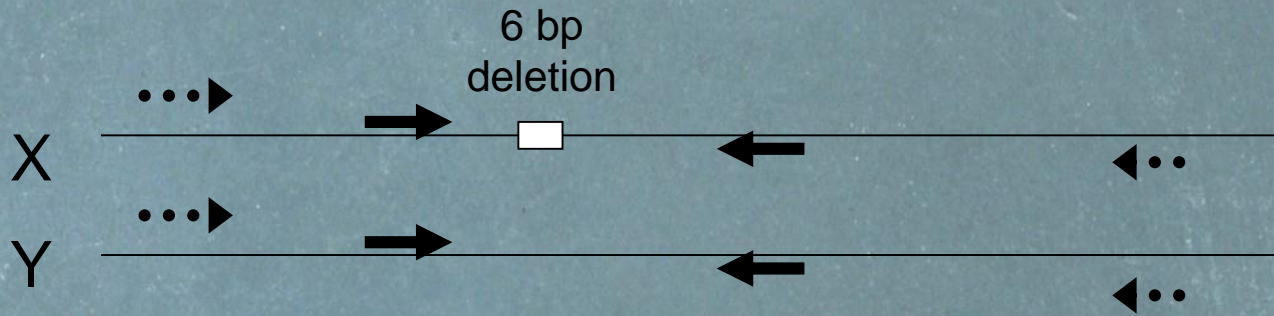
- ✓ X and Y differentiated from an ordinary autosomal pair million years ago
- ✓ Y is essentially a degraded version of X



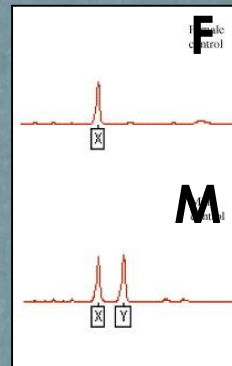
✓ The **amelogenin** gene, located in PAR 1 region of X and Y chromosomes encodes for an extracellular matrix protein found in enamel



✓ The sequence of the first intron of the amelogenin gene on X carries a 6 bp deletion compared to Y

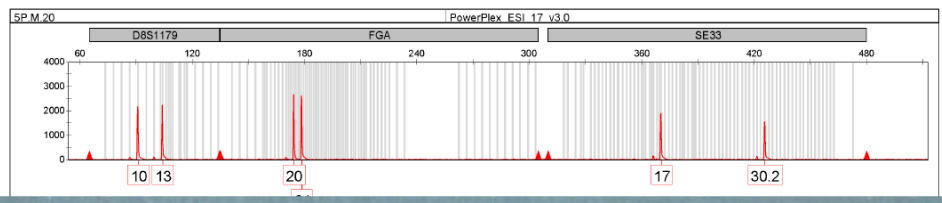
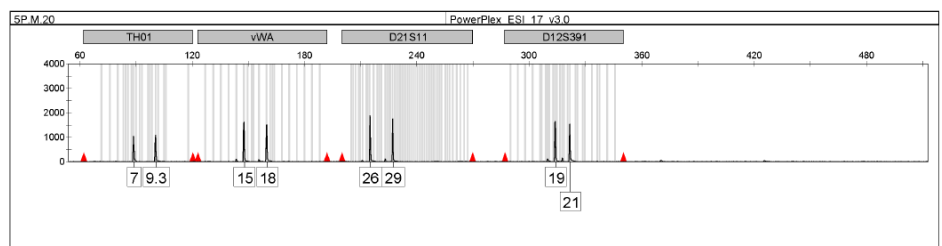
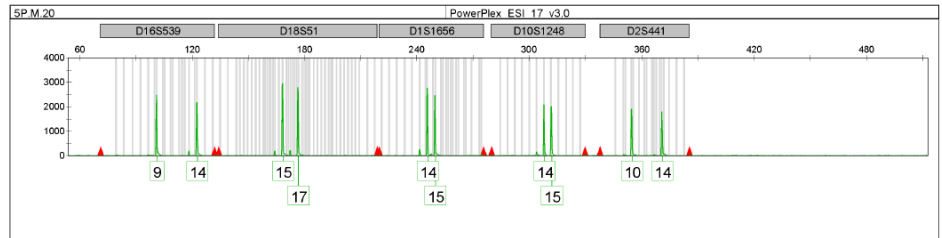
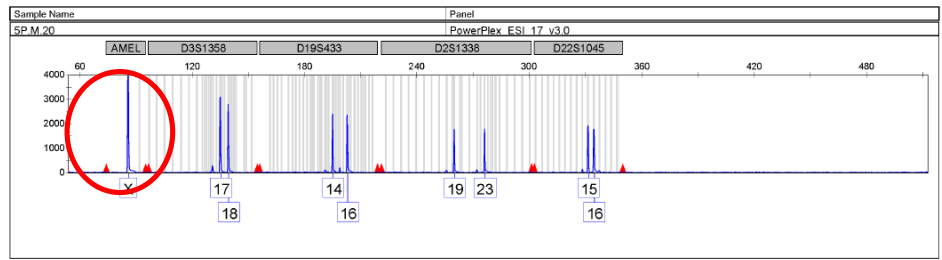


PCR produces a single peak in females, but two peaks, 6 bp apart, in males

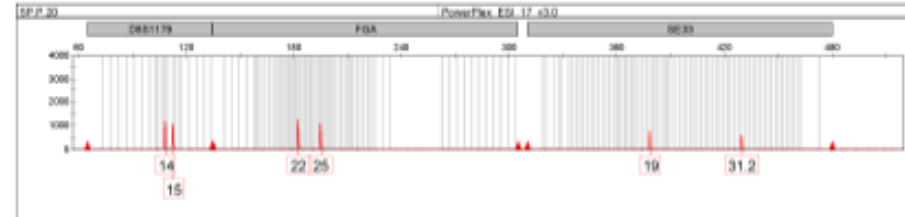
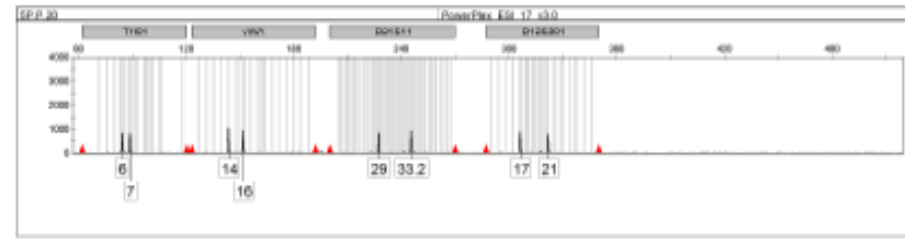
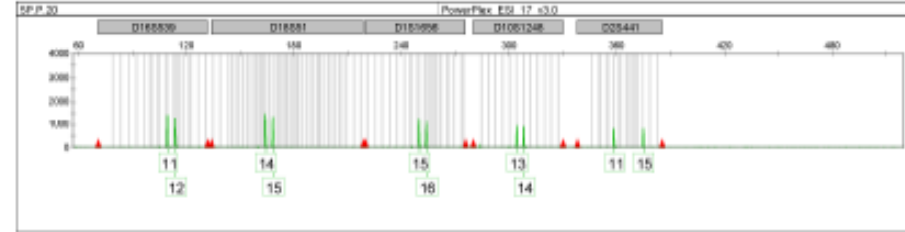
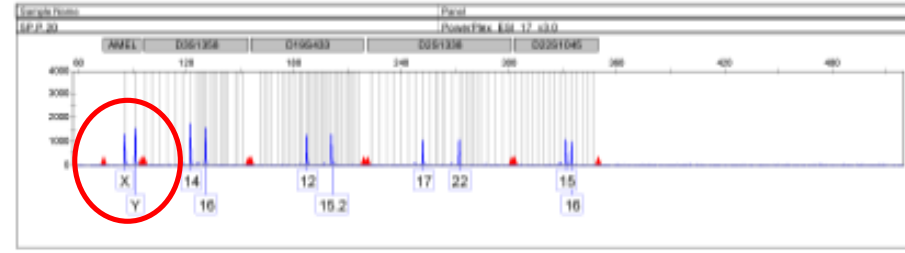


Amelogenin is always included in multiplex PCR kits for the amplification of STRs

5P-20

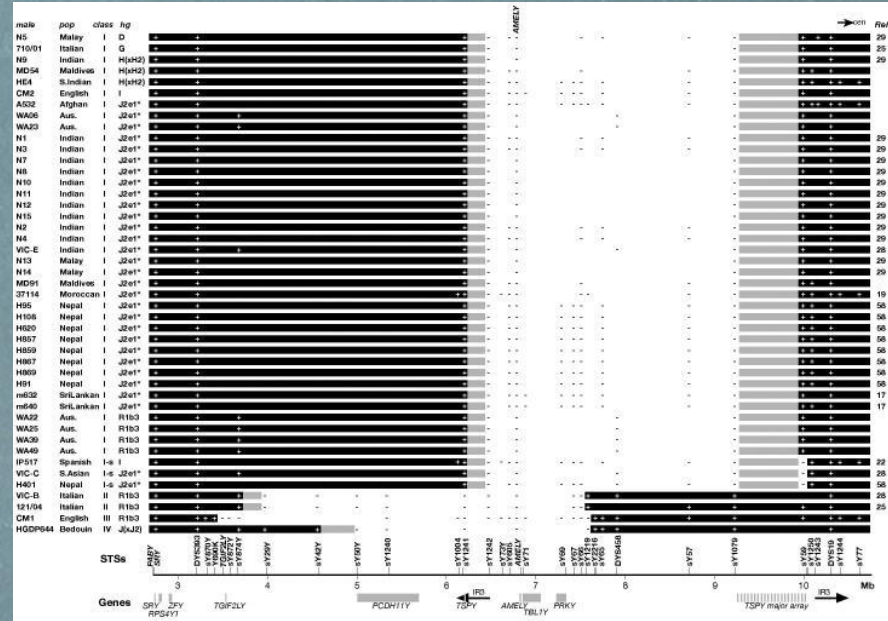


5P-20

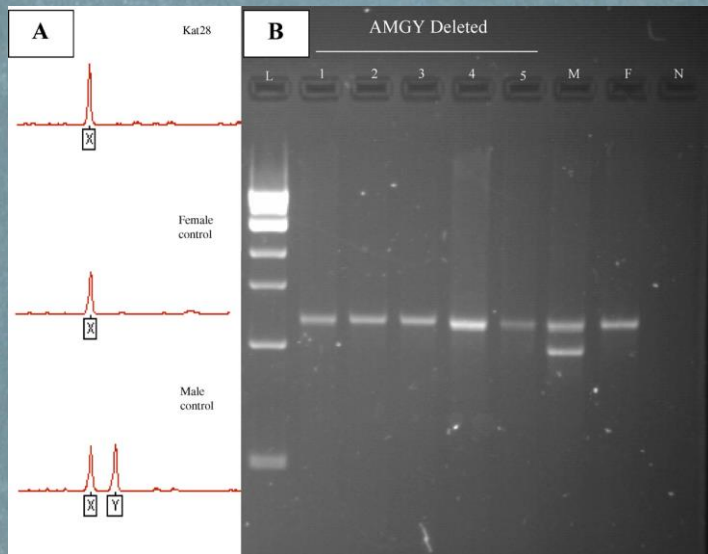


✓ A large deletion of the Y chromosome including the tract encompassing the part of the amelogenin gene used in forensic for sex-typing of stains is common in the Indian sub-continent:

- Pakistan (2%)
- India (5%)
- Nepal (6%)

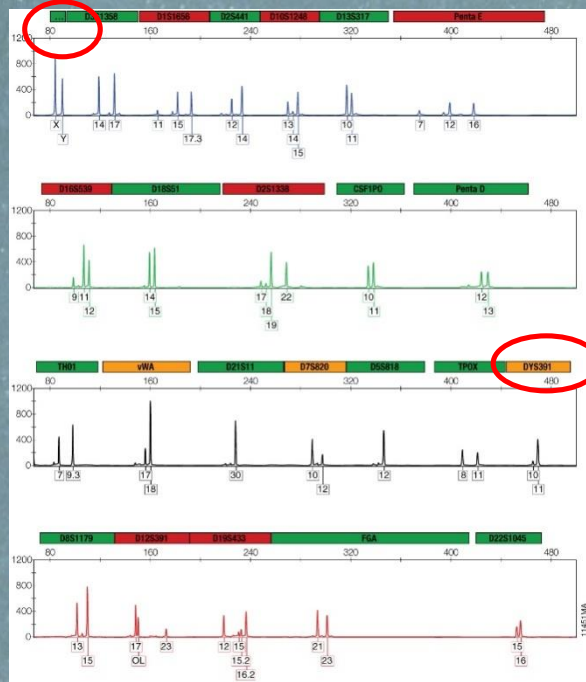


Jobling et al Hum Mol Genet 2007

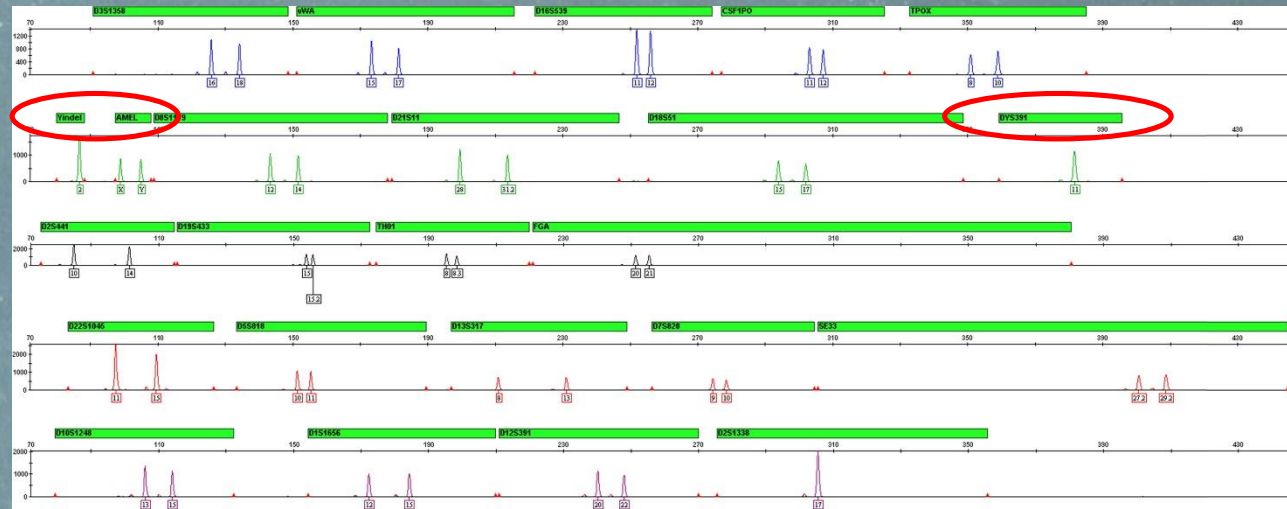


→ Wrong assignation of sex in unknown stain

Recent forensic STR kits have been modified to include additional Y specific markers, also taking advantage of upgrade from traditional 4 or 5-dye to 6-dye chemistries.



PowerPlex Fusion System (Oostdik et al. Forensic Sci Int Genet 2014)



GlobalFiler Express PCR Amplification kit (Flores et al. Forensic Sci Int Genet 2014)

MPS technology can combine >> larger number of markers compared to CE, including multiple Y and X loci

Table 1. Forensic Loci Included in ForenSeq DNA Signature Prep Kit

Feature	Number of Markers <sup>a</sup>	Amplicon Size Range (bp)
Global Autosomal STRs	27	61–467
Y-STRs	24	119–390
X-STRs	7	157–462
Identity SNPs	95	63–231
Phenotypic SNPs <sup>b</sup>	22	73–227
Biogeographical Ancestry SNPs <sup>b</sup>	56	67–200

a. SNP and STR chromosome locations can be found in the ForenSeq DNA Signature Prep Kit User Guide ([support.illumina.com/downloads/forenseq-dna-signature-prep-guide-15049528.html](http://support.illumina.com/downloads/forenseq-dna-signature-prep-guide-15049528.html)).

b. Two piSNPs used for hair/eye color are also used in the aiSNP marker set.