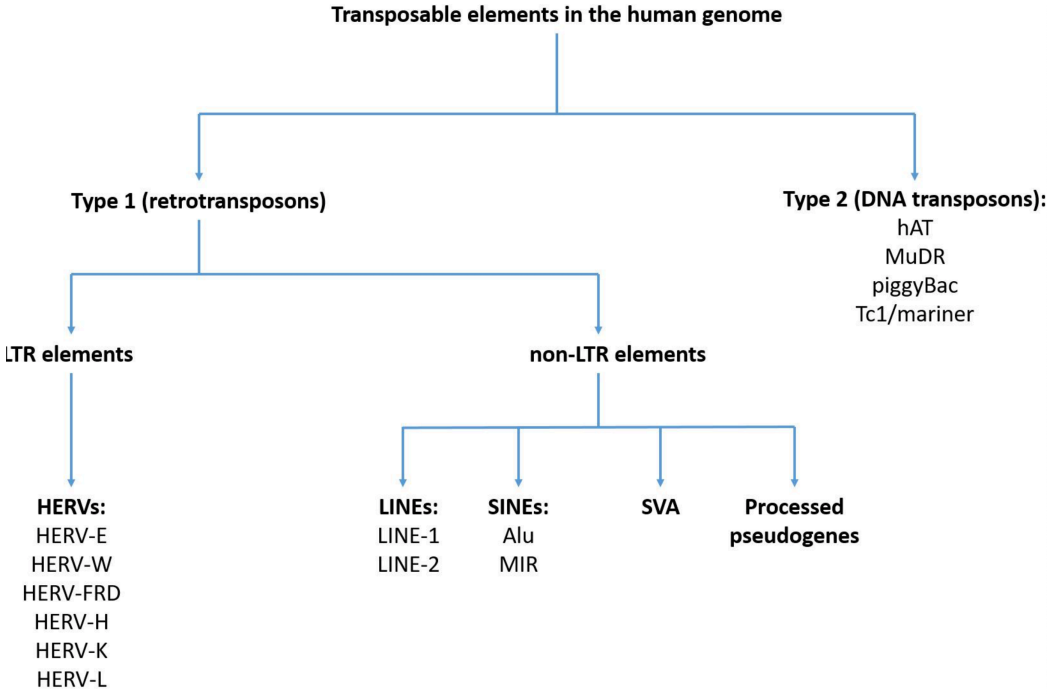
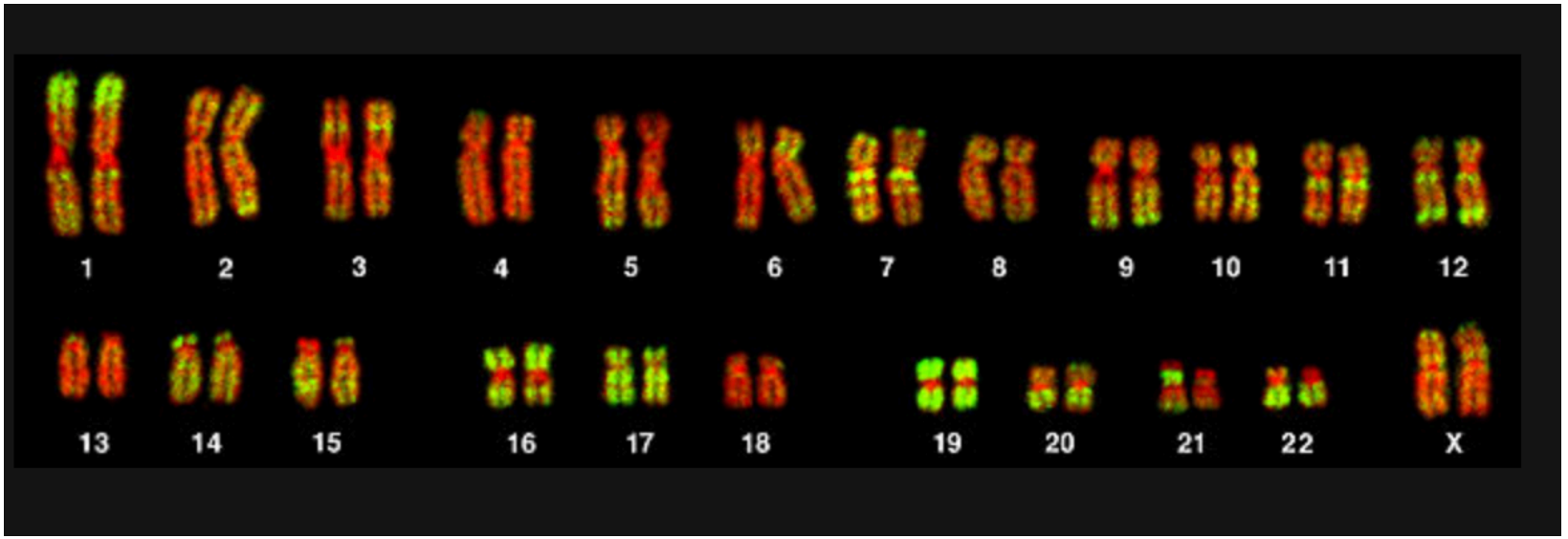
Transposable elements in the genome



ca. 50% of entire genome



DNA FISH with Alu probe

Transposable elements in the genome

- 306 base pair segment of DNA, Classified as a **SINE** (**S**hort **In**terspersed Repetitive **E**lement)
- Named for the **Alu I** restriction site within the sequence (AGCT)
- Human-specific **Alu** insertion
- Approx. 1 million **Alu** copies per haploid genome = 11% of the genome: role in genetic architecture and genetic disorders
- Intron: Found in a non-coding region of your DNA

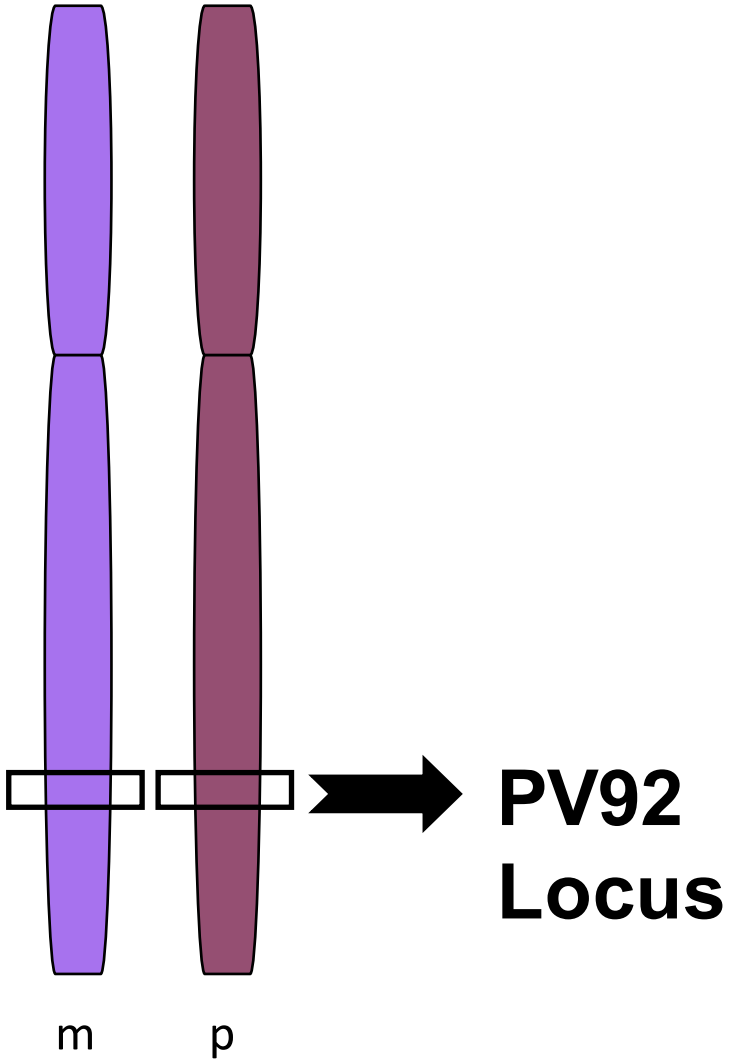
Alu seuqence insertion in PV92 locus is not **diagnostic for anydisease or disorder!**

GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCGGGCGGATCACGAGG
TCAGGAGATCGAGACCATCCCGGCTAAAACGCTGAAACCTCGTCTCTACTAAAAATACAAAAAATTAGCCG
GGCGTAGTGGCGGGCGCCTGTAGTCCC***AGCT***ACTTGGGAGGCTGAGGCAGGAGAATGGCGTGAACCCG
GGAGGCGG***AGCT***TGCAGTGAGCCGAGATCCTGCCACTGCACTCCAGCGTGGGCGACAGAGCGAGACTC
CGTCTCAAAAAAAAAAAAAAAAAAAAAAA

306 base pairs long: This sequence remains the same, no matter where it is found in the genome

Transposable elements in the genome

Chromosome 16
Homologous Chromosomes

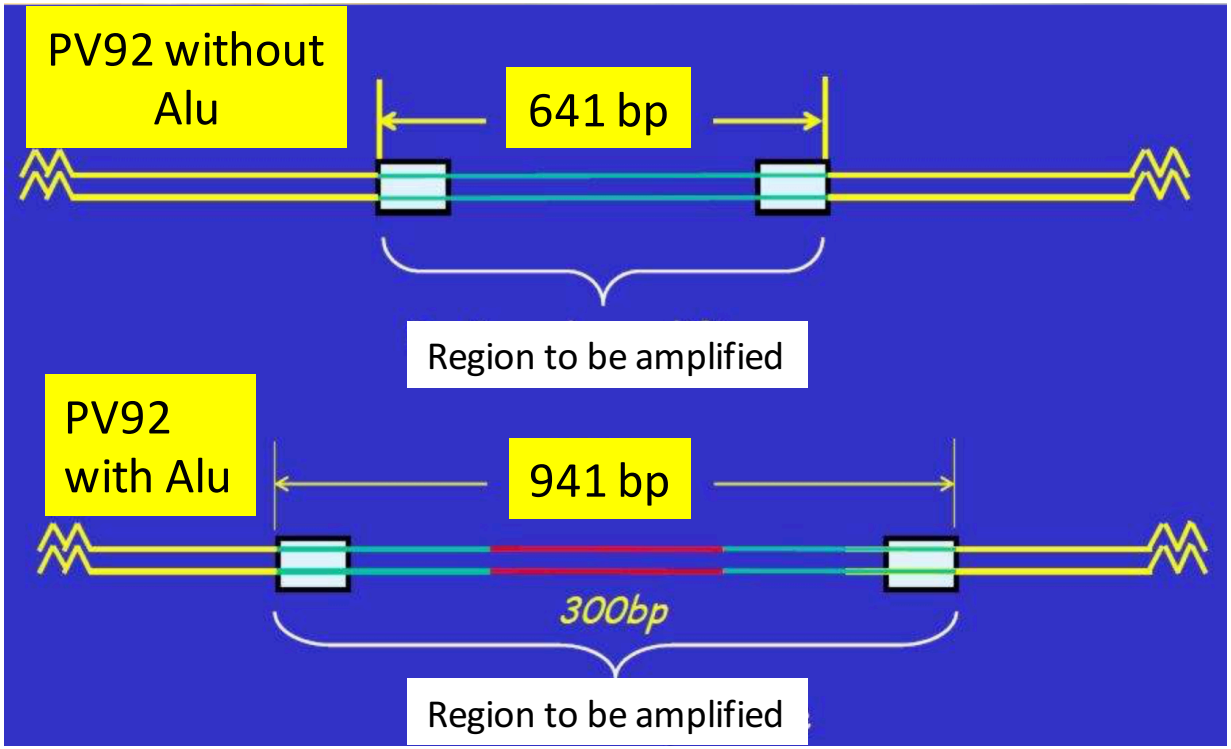


- Each gene locus has a particular form of the gene, or **allele**
- What are the possible alleles for the Alu insert at each locus?
  - +, Alu present
  - -, Alu not present
- What are the possible genotypes for the Alu insert for any given person?
  - Homozygous positive: +/+
  - Homozygous negative: -/-
  - Heterozygous: +/-

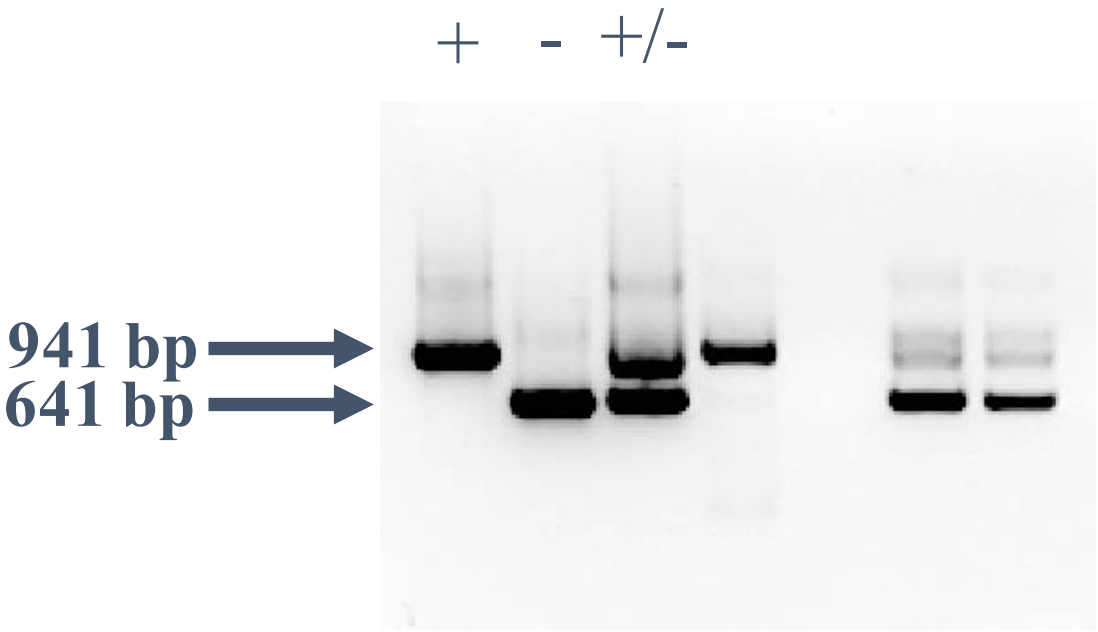
Transposable elements in the genome

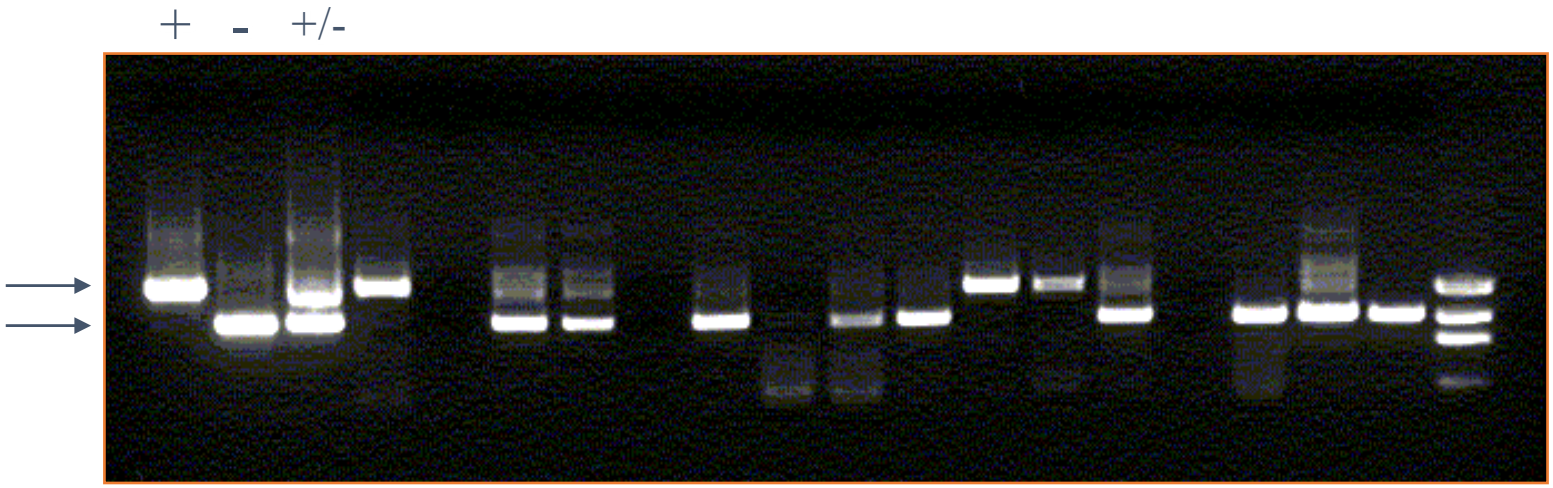
Options:

+/+ = 941 / 941 bp
+/- = 941 / 641 bp
-/- = 941 / 641 bp



Transposable elements in the genome





Evolutionary significance of PV92 Alu sequences

Facts

- Highly conserved
- Inserted in the last 1,000,000 years
- Genotypes (+/+, +/–, –/–)
- Used in population genetics, paternity analysis, and forensics
- No selective constraint on Alu sequence → passed on next generation with out selective pressure → follow distribution in large populations

Determination and analysis of *Alu* frequency in a population

- Amplify *Alu* insert from representative sample population (100 students)
- Calculate the expected allelic and genotypic frequencies
- Perform Chi-square test

Population genetics with Uni Trieste Students

Determination and analysis of *Alu* Frequency in a small student population

- Amplify *Alu* insert from representative sample population and collect data from all students
- Calculate the observed allelic and genotypic frequencies
- Perform Chi-square test

Population genetics with Uni Trieste Students

$$\text{+/+ Genotypic frequency} = \frac{\text{Number with genotype}}{\text{Population total (N)}}$$

$$\text{+ Allele frequency} = \frac{\text{Number of + alleles}}{\text{Total number of alleles}}$$

Hardy Weinberg equation to calulate genotypic frequency

$$p^2 + 2pq + q^2 = 1$$

Chi-Square Test to test whether genotypic frequencies are in genetic equilibrium in student population

$$\mathbf{X^2} = \frac{\sum \mathbf{(Observed - Expected)^2}}{\mathbf{Expected}}$$

Allelic and genotypic frequencies in different small populations

Provenienza	numero individui	alleli Alu +	alleli Alu-	omozigoti -/-	Omozigoti +/+	eterozigoti +/- e -/+
Papua Nuova Guinea entroterra	47	36,2	63,8	40,5	12,9	46,7
Papua Nuova Guinea Isole	69	23,9	76,1	57,8	5,6	36,7
Aborigeni australiani	99	15,2	84,8	71,9	2,3	25,8
Nusa Tengarras (Indonesia)	91	50	50	24,9	24,9	50,3
Moluccas (Indonesia)	49	69,4	30,6	9,2	48,0	42,9
Cinesi e vietnamiti	16	81,3	18,7	3,0	65,6	31,5
Quechua (nativi americani)	20	87,5	12,5	1,3	76,3	22,4
Arhuaco (nativi americani)	20	97,5	2,5	0,0	95,0	5
nativi Alaska (nativi americani)	62	64,5	35,5	12,4	41,4	46,2
Greci	50	25	75	56,1	6,1	37,9
Turchi	33	33,3	66,7	44,2	10,8	45,1
Caucasici nord americani	32	14,1	85,9	73,6	1,8	24,6
Zaire (pigmei)	17	35,3	64,7	41,2	11,8	47,1
Rep CentrAfricana (pigmei)	17	26,5	73,5	53,5	6,5	40,1
Nigeriani	11	9,1	90,9	82,3	0,4	17,3
Afro-Americani	31	17,7	82,3	67,5	2,9	29,7