

Polymorphism

National Human Genome Research Institute (NHGRI)

Source

National Human Genome Research Institute (NHGRI). *Polymorphism*.

Single nucleotide polymorphism (SNP)

Individual 1

Chr 2 ..CGATATTCC**T**ATCGAATGTC..
copy1 ..GCTATAAGG**A**UAGCTTACAG..

Chr 2 ..CGATATTCC**C**ATCGAATGTC..
copy2 ..GCTATAAGG**G**TAGCTTACAG..

Individual 2

Chr 2 ..CGATATTCC**C**ATCGAATGTC..
copy1 ..GCTATAAGG**G**TAGCTTACAG..

Chr 2 ..CGATATTCC**C**ATCGAATGTC..
copy2 ..GCTATAAGG**G**TAGCTTACAG..

Short tandem repeat polymorphism (STRP)

Individual 3

Repeat unit

Chr 2 ..CGATATTCC**CAGCAGCAG**ATCGAATGTC..
copy1 ..GCTATAAGG**CAGCAGCAG**TAGCTTACAG..

Chr 2 ..CGATATTCC**CAGCAGCAGCAGCAG**ATCGAATGTC..
copy2 ..GCTATAAGG**CAGCAGCAGCAGCAG**TAGCTTACAG..

Individual 4

Chr 2 ..CGATATTCC**CAGCAGCAGCAGCAG**ATCGAATGTC..
copy1 ..GCTATAAGG**CAGCAGCAGCAGCAG**TAGCTTACAG..

Chr 2 ..CGATATTCC**CAGCAGCAGCAGCAGCAGCAG**ATCGAATGTC..
copy2 ..GCTATAAGG**CAGCAGCAGCAGCAGCAGCAG**TAGCTTACAG..

Polymorphism involves one of two or more variants of a particular DNA sequence. The most common type of polymorphism involves variation at a single base pair.

Polymorphisms can also be much larger in size and involve long stretches of DNA. Called a single nucleotide polymorphism, or SNP (pronounced snip), scientists are studying how SNPs in the human genome correlate with disease, drug response, and other phenotypes.