

DNA Mutations Explained



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Setting the Standard for Quality DNA Identification

The human genome is very large, containing more than 3.2 billion bases, or letters. Despite its size, over 99.9% of the DNA in all unrelated people in the world is exactly the same. Consequently, the vast differences observed in the human race are created from the differences in only 0.1% of DNA. These differences are caused by mutations that have occurred gradually over the millennia.

As with any region of DNA, the STR genetic markers used in relationship and paternity testing are subject to DNA mutations, also called genetic inconsistencies. They are naturally occurring mismatches between a child and parent and can occur when a sperm cell is being made or when a cell divides. The majority of STR mutations involve the gain or loss of a single unit. Thus, a maternal or paternal genetic marker with a 14 would show as a 13 or 15 in the next generation following a mutational event. These mutations can occur in either the mother or the father, but appear to be more frequent in the father. On average, the genetic markers used in relationship testing mutate at an approximate rate of 0.13%.

The colored images below illustrate how a DNA mutation can be passed down from the father to the child. As you can see, the mother passes one of her chromosomes to the child, and the father passes a mutated chromosome that increased by one unit.

Mother		Child		Father	
Allele A =10	Allele B =10	Allele A =10	Allele B =15	Allele A =14	Allele B =14

The frequencies of these naturally occurring mismatches are factored into the final combined paternity index. Due to the nature of these calculations, the statistical value of the result is significantly reduced; therefore some degree of extended testing is typically required to obtain a conclusive result. This usually involves testing the mother if she was not included in the original test. Alternatively, additional autosomal markers can be tested, or Y chromosome markers can be tested if the child is male and if the potential alleged fathers are not paternally related.



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