

Quick reference - simple examples description of sequence changes

Last modified August, 2015

Since references to WWW-sites are not yet acknowledged as citations, please mention [den Dunnen JT and Antonarakis SE \(2000\). Hum.Mutat. 15:7-12](#) when referring to these pages.

Simple examples

For detailed descriptions and more extensive examples see *Examples at [DNA](#), [RNA](#) or [protein](#) level*. For an explanation of the characters used in the descriptions [see Characters used](#). For mitochondrial DNA variants [see Reference Sequence](#).

Type of change	DNA (examples shown relate a to coding DNA reference sequence)	RNA (report only when RNA was analysed)	protein (NOTE, change deduced only !)
substitution	c.-7A>C		
	c.3G>T		
	c.88+2T>G		
	c.89-1G>T		p.Trp26Cys (p.W26C)
	c.*23T>C	r.67g>u	p.Trp26* (p.W26*)
	NM_004006.1:c.3G>T		

	GJB6:c.3G>T rs2306220:A>G		
deletion	c.13del (c.13delT) c.13_16del c.120_123+4del c.13-?_300+?del	r.13del (r.13delu) r.13_16del r.13_300del	p.Gly4del (p.G4del) p.Gly4_Gln6del (p.G4_Q6del) p.Gly4Valfs*14 (p.Gly4fs)
duplication	c.13dup (c.13dupT) c.92_94dup (c.92_94dupGAC)	r.13dup (r.13dupu) r.92_94dup (r.92_94dupgac)	p.Gly4dup (p.G4dup) p.Gly4_Gln6dup (p.G4_Q6dup)
insertion	c.51_52insT c.51_52insGAGA	r.51_52insu r.51_52insgaga	p.Lys2_Leu3insGlnSer (p.K2_L3insQS)
variability short sequence repeats	c.162CAG(12_34) c.123+74_123+75(3_6) g.958A(19_23)	r.162cag(12_34)	p.Gln54(12_34) (p.Q(12_34))
inversion	c.77_80inv	r.77_80inv	-
complex	c.112_117delinsTG (c.112_117delinsTG)	r.112_117delinsug (r.112_117delinsug)	p.Cys28_Lys29delinsTrp (p.C28_K29delinsW)
two changes in one allele	c.[76C>T; 83G>C]	r.[r.76C>T; r.83G>C]	p.[Trp13*; Ala43Pro]
two changes in one individual, alleles unknown	c.[76C>T(;)181T>C]	r.[76C>T(;) 181T>C]	p.[Trp13*(;)Trp61Arg]
recessive disease (changes in	c.[76C>T];[87G>A] c.[76C>T];[?]	r.[76c>u];[87g>a] r.[76c>u];[?]	p.[Trp13*];[Cys28Arg] p.[Trp13*];[?]

<i>different alleles)</i>	c.[76C>T];[=]	r.[76c>u];[=]	p.[Trp13*];[=]
recessive disease (changes in different genes)	GJB2:c.[76C>T] ; GJB6:c. [87G>A]	GJB2:r.[76c>u] ; GJB6:r.[87g>a]	GJB2:p.[Trp13*] ; GJB6:p.[Cys28Arg]
exact location unknown (see Uncertain)	c.(67_70)insG c.(165_253)del11 c.88-?_923+?del c.(?-244)_32+?del	r.(67_70)insg r.(165_253)del11 r.88_923del r.0?	p.Gly23fs p.fs p.Thr29fs*16 p.0?
frame shift (protein only, see Examples)	-	-	p.Gly44Valfs*14 (p.Gly4fs) p.Leu30Serfs*3 (p.Leu30fs)

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