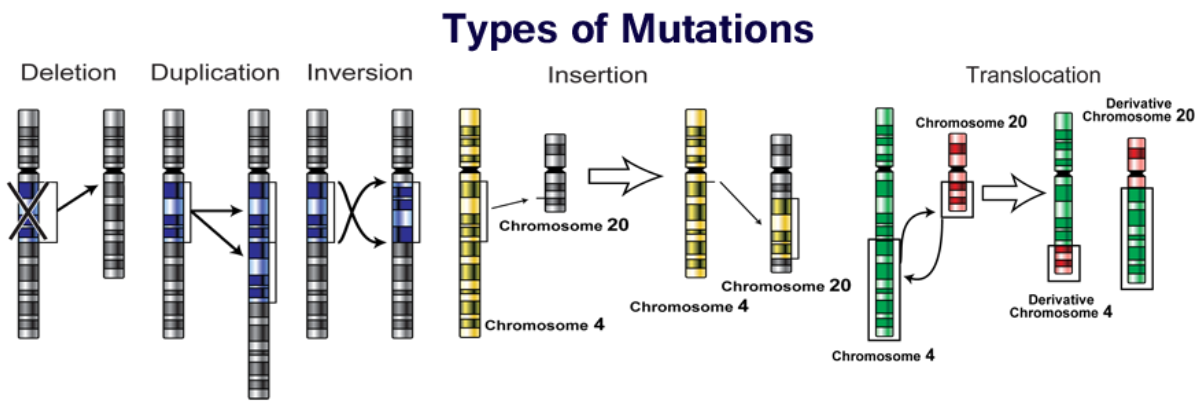
CYTOGENETIC TERMINOLOGY

Common punctuation seen in a cytogenetic report.

Description	Symbol	Significance
Bracket, angle	<n >	Denotes ploidy level (the # of complete sets of chromosomes in a cell)
Bracket, square	[ ]	Denotes number of cells in a cell line/clone, # of metaphases examined
Colon, single	:	Chromosomal break in the detailed system
Colon, double	::	Chromosomal break and reunion in the detailed system & fusion genes
Comma	,	Separates chromosome numbers and chromosome abnormalities
Minus sign	-	Loss or decrease in length
Multiply sign	X	Multiple copies of rearranged chromosomes or number of copies
Parenthesis	( )	Surround structurally altered chromosomes and breakpoints, or genes
Plus sign	+	Additional normal or abnormal chromosome, or increase in length
Semicolon	;	Separates altered chromosome and breakpoints in structural rearrangements involving more than one chromosome.
Slant	/	Separates cell lines, clones or contiguous probes
Greater than	>	Substitution

Common abbreviations seen on a cytogenetic report.

Aberration	Example	Description
add	add(1)(q21)	Additional material attached to a chromosome region or band.
del	del(7)(q22q31)	Deletion or loss of chromosome material.
der	der(5)inv(5)	A structurally rearranged chromosome from either a rearrangement involving 2 or more chromosome or by multiple aberrations within a single chromosome.
dic	45,XY,dic(13;14)	One chromosome replaces 2 normal chromosomes resulting in 2 centromeres. The 2 chromosome segments fuse, resulting in the loss of the acentric fragments.
dup	dup(1)(q21q32)	Part of a chromosome is repeated.
ins	ins(5)(p13q31q15)	Addition of material from another chromosome.
inv	inv(9)(p13q15)	Part of the chromosome is inverted within the chromosome.
rec	rec(6)	Recombinant chromosome due to meiotic crossing-over.
t	t(8;9;22)(p21;q34.1;q11.2)	Material between 2 different chromosomes is exchanged.



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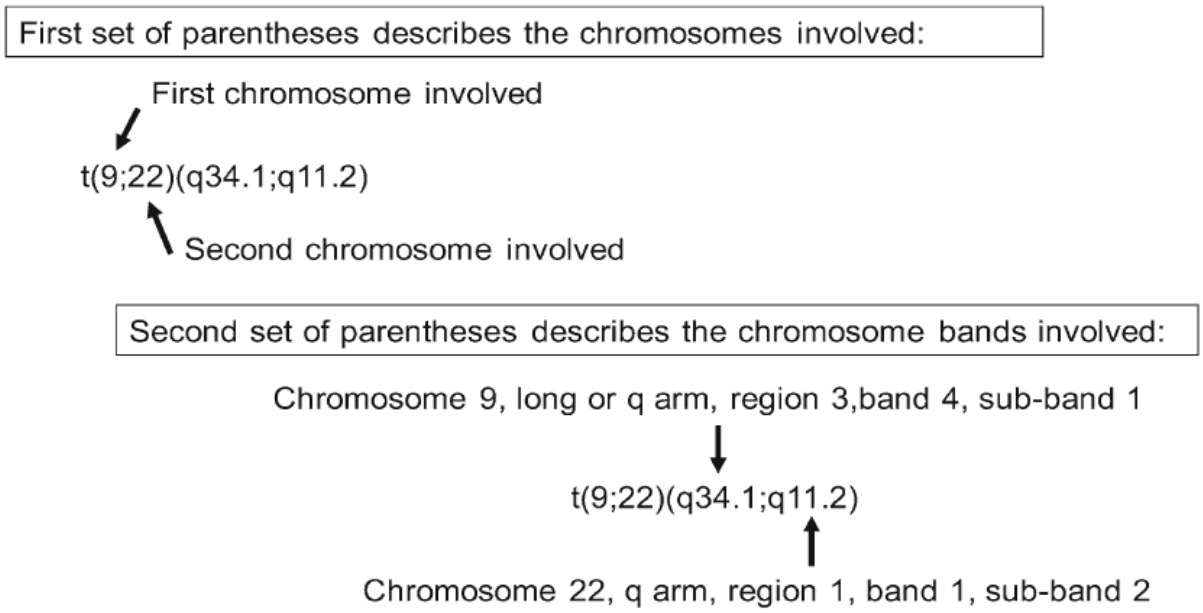

GENE	Location	Testing	Description
TP53	17p13.1	FISH, NGS, PCR	Regulation of cell division, acts as a tumor suppressor.
IgVH	14q32.33	NGS, PCR	Involved in the production of antibodies by B cells.
DLEU2/MIR15A/MIR16-1 RB1	13q14	FISH,Microarray Karyotyping,PCR	Tumor suppressor Regulating cell cycle progression and genomic stability
Trisomy 12	+12	FISH, Karyotyping	There is an extra copy of chromosome 12; Cell adhesion and migration
ATM	11q22-q23	FISH,Microarray Karyotyping	DNA repair and cell cycle control
FGFR3	4p16.3	FISH,Microarray Karyotyping,PCR	t(4;14), associated with aggressive disease
CCND1	11q13	FISH,Microarray Karyotyping,PCR	Regulation of cell cycle. t(11;14) is a favorable prognosis
MAF	16q23	FISH,Microarray Karyotyping,PCR	t(14;16), linked to poor prognosis
MAFB	20q12	FISH,Microarray Karyotyping,PCR	t(14;20), linked to poor prognosis
BCR::ABL	t(9;22)	FISH, Karyotyping, PCR	Philadelphia chromosome encodes a protein with abnormal tyrosine kinase activity, which leads to uncontrolled cell division
MSH2, MSH6, MLH1, PMS2	3, 2 and 7	NGS, IHC, PCR	Mismatch repair (MMR) system, DNA repair mechanism that maintains genetic stability.
MYC	8q24.21	FISH, IHC, PCR, NGS	Cell cycle progression, apoptosis & cell transformation

This list is not all inclusive