

# From SNPs to pathways: Integration of functional effect of sequence variations on models of cell signalling pathways

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## Glossary

**Variation:** any kind of short range sequence variation in the nucleotide sequence of the genome. Among these we can mention SNPs and insertion-deletion variants of a small number of nucleotides, Alu sequences and other types of variations collected in dbSNP database .

**SNPs:** single nucleotide polymorphisms or SNPs are DNA sequence variations in which a single nucleotide (A, G, C or T) is altered. SNPs are also referred as polymorphisms, natural variants, or common variants because they have a minor allele frequency in the population of at least 1 %. In contrast, rare variants have a minor allele frequency of less than 1 % . SNPs can either be neutral or have an effect on gene or protein function.

**Mutation:** term used to refer to rare variants (minor allele frequency less than 1 % in the population). These variants often cause disease and segregate within families, and affect conserved residues in the protein sequence. Also used to refer to modified residues in a sequence after the experimental procedure of mutagenesis.

**Mutagenesis:** experimental procedure aimed at altering the protein sequence at residues suspected to be critical for the function of the protein.

## Identifiers and names of proteins used in the study

Protein name (as used in this study)	UniProt id	Protein name according to UniProt
SOS1	Q07889	Son of sevenless homolog 1
RAS  (generic term for HRas, KRas, NRas)	P01112	GTPase Hras
	P01116	GTPase Kras
	P01111	GTPase Nras
ERK  (generic term for ERK1, ERK2)	P27361	Mitogen-activated protein kinase 3  (ERK1)
	P28382	Mitogen-activated protein kinase 1  (ERK2)
EGFR or ErbB1	P00533	Epidermal growth factor receptor
Grb2	P62993	Growth factor receptor-bound protein  2
EGF	P01133	Pro-epidermal growth factor
MEK  (generic term for MEK1, MEK2)	Q02750	Dual specificity mitogen-activated  protein kinase 1 (MEK1)
	P36507	Dual specificity mitgen-activated  protein kinase 2 (MEK2)
MEKstar (activated MEK1)	Q02750	Dual specificity mitogen-activated  protein kinase 1 (MEK1)
RAF	P04049	RAF proto-oncogene  serine/threonine-protein kinase
Akt	Q9Y243	RAC-gamma serine/threonine-protein  kinase
Aktstar (activated Akt)	Q9Y243	RAC-gamma serine/threonine-protein  kinase
ErbB2 or HER2/NEU	P04626	Receptor tyrosine-protein kinase

		erbB-2
ErbB3	P21860	Receptor tyrosine-protein kinase erbB-3
ErbB4	Q15303	Receptor tyrosine-protein kinase erbB-4
HRG	Q02297	Pro-neuregulin-1, membrane-bound isoform (heregulin)
Gab1	Q13480	GRB2-associated-binding protein 1
PTP1-B	P18031	Tyrosine-protein phosphatase non- receptor type 1