Human genes associated with disease by missense mutations:

Parkinson's disease: Alpha-syn nuclein - G209A

Rett syndrome: MECP2
R133C
F155S
T158M
R106W

Tangier disease: ABC Transporter1
W530S
N875S
A877V

Early onset Parkinson's disease: PINK1
G309D

Desmin-related myopathy: alpha-B-crystallin chaperone
R120G

Hemophilia C: Factor VIII
C1 domain
22 Q2087E
22 R2090C
23 R2150C
23 R2163C
23 M2164R
C2 domain
24 S2173I
24 A2201P
25 V2232A
26 P2300L
26 R2304C
26 R2304G
26 R2307Q
26 R2320T

Table 1 • *FLT4* mutations linked to hereditary lymphoedema

Mutation	Bp of cDNA	Codon change	Location	Effects
P641S	1,921	Pro (CCC)→Ser (TCC)	extracellular VI Ig-homology domain	no effect (polymorphism)
G857R	2,569	Gly (GGG)→Arg (AGG)	intracellular I TK domain (ATP-binding motif)	TK negat.
R1041P	3,122	Arg (CGG)→Pro (CCG)	intracellular II TK domain	TK negat.
L1044P	3,131	Leu (CTG)→Pro (CCG)	intracellular II TK domain	TK negat.
P1114L	3,341	Pro (CCG)→Leu (CTG)	intracellular II TK domain	TK negat.

Mutation of the Stargardt Disease Gene (ABCR) in Age-Related Macular Degeneration

Table 1. Prevalence of AMD-associated variants in *ABCR* (*23*). NA, data not available.

Mutation	AMD (n =167)	STGD (n = 98)	General population (n = 220)
E471K	2 (1.2%)	NA	0 (0%)
R1129L	1 (0.6%)	0 (0%)*	0 (0%)
T1428M	1 (0.6%)	0 (0%)	0 (0%)
R1517S	1 (0.6%)	0 (0%)	0 (0%)
I1562T	2 (1.2%)	0 (0%)	0 (0%)
G1578R	1 (0.6%)	0 (0%)	0 (0%)
5196+1G →A	1 (0.6%)	0 (0%)	0 (0%)
R1898H	1 (0.6%)	4 (4%)	0 (0%)
G1961E	6 (3.6%)	8 (8%)	0 (0%)
L1970F	1 (0.6%)	0 (0%)	0 (0%)
6519Δ11bp	1 (0.6%)†	1 (1%)†	0 (0%)
D2177N	7 (4.2%)	0 (0%)	1 (0.45%)
6568ΔC	1 (0.6%)	0 (0%)	0 (0%)
Totals	26 (16%)	13 (13%)	1 (0.45%)

*A substitution to a different amino acid (R1129C) was detected in one STGD1 patient. †The two individuals with this variant are related (Fig. 2).

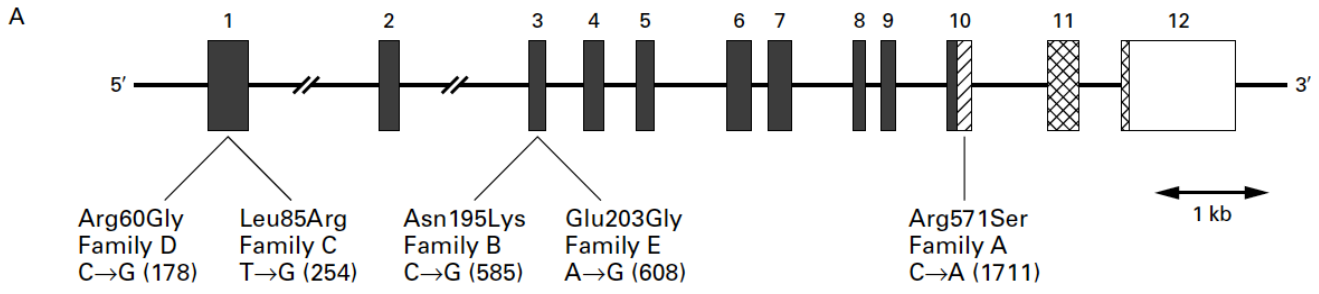
Mutations of the BRAF gene in human cancer

Table 1 *BRAF* mutations in human c

BRAF mutations

Nucleotide	Amino acid
G1388A	G463E
G1388T	G463V
G1394C	G465A
G1394A	G465E
G1394T	G465V
G1403C	G468A
G1403A	G468E
G1753A	E585K
T1782G	F594L
G1783C	G595R
C1786G	L596V
T1787G	L596R
T1796A	V599E
TG1796-97AT	V599D
	Total
No. samples screened	
Per cent	

MISSENSE MUTATIONS IN THE ROD DOMAIN OF THE LAMIN A/C GENE AS CAUSES OF DILATED CARDIOMYOPATHY AND CONDUCTION-SYSTEM DISEASE



Missense mutations in the most ancient residues of the PAX6 paired domain underlie a spectrum of human congenital eye malformations