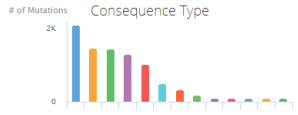
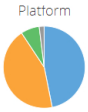
ADVANCED SEARCH

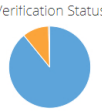
Donors | Genes | Mutations

Share | Show PQL | Clinical Significance IN (Uncertain significance , Likely pathogenic , Pathogenic ...)

Donors 9,906 | Genes 1,580 | Mutations 3,145







Mutations 3,145 | Mutation Occurrences 16,875

Somatic Mutations

Genome Viewer | Save/Edit Mutation Results

Showing **1** - **10** of **3,145** mutations

ID	DNA change	Type	Consequences	Clinical Significance	# Donors affected: Total	# Donors affected: Across all Projects
MU62030	chr7:g.140453136A>T	single base substitution	Missense: *BRAF* V207E, V600E 3 UTR: *BRAF* Exon: *BRAF*	Pathogenic	814 / 9,906 (8.22%)	814 / 19,729 (4.13%)
MU37643	chr12:g.25398284C>T	single base substitution	Missense: *KRAS* G12D	Pathogenic	533 / 9,906 (5.38%)	533 / 19,729 (2.70%)
MU12519	chr12:g.25398284C>A	single base substitution	Missense: *KRAS* G12V	Pathogenic	401 / 9,906 (4.05%)	401 / 19,729 (2.03%)
MU866	chr2:g.209113112C>T	single base substitution	Missense: *IDH1* R132H Exon: *IDH1* Downstream: *IDH1*	Pathogenic/Likely pathogenic	400 / 9,906 (4.04%)	400 / 19,729 (2.03%)
MU4468	chr3:g.178952085A>G	single base substitution	Missense: *PIK3CA* H1047R Downstream: *RP11-245C23.3*	Pathogenic	370 / 9,906 (3.74%)	370 / 19,729 (1.88%)
MU5219	chr3:g.178936091G>A	single base substitution	Missense: *PIK3CA* E545K Upstream: *PIK3CA*	Pathogenic/Likely pathogenic	354 / 9,906 (3.57%)	354 / 19,729 (1.79%)
MU7870	chr17:g.7578406C>T	single base substitution	Missense: *TP53* R82H, R175H, R43H Upstream: *TP53* Exon: *TP53* Downstream: *TP53*	Pathogenic	268 / 9,906 (2.71%)	268 / 19,729 (1.36%)
MU5136	chr3:g.178936082G>A	single base substitution	Missense: *PIK3CA* E542K Upstream: *PIK3CA*	Pathogenic/Likely pathogenic	238 / 9,906 (2.40%)	238 / 19,729 (1.21%)
MU30748	chr17:g.7577538C>T	single base substitution	Missense: *TP53* R116Q, R155Q, R248Q Upstream: *TP53* Exon: *TP53* Downstream: *TP53*	Pathogenic/Likely pathogenic	192 / 9,906 (1.94%)	192 / 19,729 (0.97%)
MU5286	chr17:g.7577121G>A	single base substitution	Missense: *TP53* R141C, R273C Upstream: *TP53* Exon: *TP53* Downstream: *TP53* Intron: *TP53*	Conflicting interpretations of pathogenicity	189 / 9,906 (1.91%)	189 / 19,729 (0.96%)

Showing 10 rows

<<< < 1 2 3 4 5 > >>>

Mutation

e.g. MU123

Upload Mutation Set

Consequence Type

- Frameshift 26
- Missense 2,122
- Start Lost 9
- Initiator Codon 1
- Stop Gained 348

Select all — 15 more

Type

- Substitution 3,095
- Deletion 46
- MSub 4

Select all

Functional Impact

- High 1,603
- Low 936
- Unknown 2,825

Select all

Clinical Significance

- Uncertain signific... 844
- Likely pathogenic 805
- Pathogenic 490
- Not provided 463
- Benign 153
- Pathogenic/Likel... 121
- Likely benign 79
- Other 65
- Conflicting interp... 55
- Association 20
- Drug response 15
- Benign/Likely be... 8
- No interpretation... 5
- Risk factor 5
- Pathogenic/Likel... 4
- Pathogenic/Likel... 4
- Likely pathogenic... 2
- Likely pathogenic... 2
- Benign, other 1
- Conflicting interp... 1
- Likely pathogenic... 1
- Pathogenic/Likel... 1
- Association not f... 1
- No Data 81,779,443

Select all / none

Clinical Evidence

- D - Preclinical 82
- C - Case 65
- B - Clinical 54
- E - Inferential 6
- A - Validated 3

Select all — 1 more

Study

Platform

Analysis Type

Verification Status

Chromosome

Location

e.g. chr12:43566-3457633

Search