

Quick Start: SNV (genomic)

Reference is **hg18**
event is **SNV**

position is **7518131**
mutant allele is **A**

The screenshot shows a web form for variant analysis. It is divided into two main sections: 'Reference sequence' and 'Variant type'.
1. Reference sequence: A dropdown menu is set to 'NCBI36 (hg18)'.
2. Variant type: A dropdown menu is set to 'SNV (single nucleotide variation)'.
3. Start position: A text input field contains '7519131'.
4. Mutant allele: A text input field contains 'A'.
5. A green button at the bottom is labeled 'Start the analysis'.
Other fields include 'End position' (7519131), 'Wild type allele (coding strand)' (G), 'Strand polarity' (Positive), and 'Sample ID (optional)' (test).

1 Select the reference sequence

A dropdown menu showing the following options:
✓ NCBI36 (hg18)
GRCh37 (hg19)
GRCh38 (hg38)
NM_000546.5 (RNA)

→ **hg18**

2 Select SNV as a variation type (default option)

3 Enter the position → **7519131**

4 Enter the mutant allele → **A**

5 Push Start



The results page displays the following information:

Reference	Start	End	Wild type	Mutant	Type
HG18	7519131	7519131	G	A	SNP

Nomenclature

Mutation in HGVS format
NG_017013.2:g.17463G>A

NCBI36 (hg18)
chr17:g.7519131G>A

GRCh37 (hg19)
chr17:g.7578406G>A

GRCh38 (hg38)
chr17:g.7675088G>A

Description

Records in database
3428

Classification
Missense_Mutation

Comment
Exonic mutation

Frequency
This single nucleotide variant is very frequent

Full description according to LRG

Affected transcripts

- LRG_32111:c.524G>A
- LRG_32112:c.524G>A
- LRG_32113:c.524G>A
- LRG_32114:c.524G>A
- LRG_32115:c.128G>A
- LRG_32116:c.128G>A
- LRG_32117:c.128G>A
- LRG_32118:c.407G>A

Affected proteins

- LRG_321p1:p.R175H
- LRG_321p3:p.R175H
- LRG_321p4:p.R175H
- LRG_321p5:p.R43H
- LRG_321p6:p.R43H
- LRG_321p7:p.R43H
- LRG_321p8:p.R136H
- LRG_321p9:p.R136H
- LRG_321p10:p.R136H
- LRG_321p11:p.R16H
- LRG_321p12:p.R16H
- LRG_321p12:p.R16H

TP53 proteins

- TP53 alpha
p.R175H
- TP53 beta
p.R175H
- TP53 gamma
p.R175H
- TP53 domain
HCD III - DNA binding
- Structural motif
Loop L2
- Post-translational modifications
-

Comments

- Activity
Inactive
- Isoforms
This substitution targets 12 TP53 isoforms
- Prediction
Damaging

Quick Start: SNV (cDNA)

Reference is **NM_000546.5 (full Length RNA)** position is **524**
event is **SNV** mutant allele is **A**

The screenshot shows a form for entering variant information. It is divided into two main columns: 'Reference sequence' and 'Variant type'.
1. Reference sequence: A dropdown menu showing 'NM_000546.5 (RNA)'.
2. Variant type: A dropdown menu showing 'SNV (single nucleotide variation)'.
3. Start position: A text input field containing '524'.
4. End position: A text input field containing '524'.
5. Wild type allele (coding strand): A text input field containing 'G'.
6. Mutant allele (coding strand): A text input field containing 'A'.
7. Strand polarity (leave positive if you have no information): A dropdown menu showing 'Positive'.
8. Sample ID (optional): A text input field containing 'Sample ID'.
9. A green 'Start the analysis' button is at the bottom.

1 Select the reference sequence

- NCBI36 (hg18)
- GRCh37 (hg19)
- GRCh38 (hg38)
- ✓ NM_000546.5 (RNA)

→ **NM_000546.5**

2 Select **SNV** as a variation type (default option)

3 Enter the position → **524**

4 Enter the mutant allele → **A**

5 Push Start



Input data					
Reference	Start	End	Wild type	Mutant	Type
NM_000546.5	524	524	G	A	SNP
Nomenclature		Full description according to LRG		TP53 proteins	
Mutation in HGVS format NG_017013.2:g.17463G>A NCBI36 (hg18) chr17:g.7519131G>A GRCh37 (hg19) chr17:g.7578406G>A GRCh38 (hg38) chr17:g.7675088G>A		Affected transcripts LRG_321t1:c.524G>A LRG_321t2:c.524G>A LRG_321t3:c.524G>A LRG_321t4:c.524G>A LRG_321t5:c.128G>A LRG_321t6:c.128G>A LRG_321t7:c.128G>A LRG_321t8:c.407G>A Affected proteins LRG_321p1:p.R175H LRG_321p3:p.R175H LRG_321p4:p.R175H LRG_321p5:p.R43H LRG_321p6:p.R43H LRG_321p7:p.R43H LRG_321p8:p.R136H LRG_321p9:p.R136H LRG_321p10:p.R136H LRG_321p11:p.R16H LRG_321p12:p.R16H		TP53 alpha p.R175H TP53 beta p.R175H TP53 gamma p.R175H TP53 domain HCD III - DNA binding Structural motif Loop L2 Post-translational modifications -	
Description				Comments	
Records in database 3428 Classification Missense_Mutation Comment Exonic mutation Frequency This single nucleotide variant is very frequent				Activity Inactive Isoforms This substitution targets 12 TP53 isoforms Prediction Damaging	

Quick Start: deletion (genomic)

Reference is **hg19**

position is **7578223** to **7578222**

event is **Del of 2 nucleotides**

! Nomenclature of the TP53 gene goes backward

The screenshot shows a variant caller interface with the following fields and callouts:

- 1** Reference sequence: GRCh37 (hg19)
- 2** Variant type: Deletion
- 3** Start position (larger than end position): 7578223
- 4** End position: 7578222
- 5** Wild type allele (coding strand): GA
- Mutant allele (coding strand): e.g., ATGC...
- Strand polarity (leave positive if you have no information): Positive
- Sample ID (optional): Sample ID
- 6** Start the analysis

1 Select the reference sequence

NCBI36 (hg18)
✓ GRCh37 (hg19)
GRCh38 (hg38)
NM_000546.5 (RNA)

→ **hg19**

2 Select Deletion as a variation type

3 Enter the start position of the deletion → 7578223

4 Enter the end position of the deletion → 7578222

5 Deleted nucleotides are automatically displayed

6 Push Start →

Reference	Start	End	Wild type	Mutant	Type
HG18	7519131	7519131	G	A	SNP

Nomenclature	Full description according to LRG	TP53 proteins
Mutation in HGVS format NG_017013.2:g.17463G>A NCBI36 (hg18) chr17:g.7519131G>A GRCh37 (hg19) chr17:g.7578406G>A GRCh38 (hg38) chr17:g.7675088G>A	Affected transcripts LRG_321t1:c.524G>A LRG_321t2:c.524G>A LRG_321t3:c.524G>A LRG_321t4:c.524G>A LRG_321t5:c.128G>A LRG_321t6:c.128G>A LRG_321t7:c.128G>A LRG_321t8:c.407G>A Affected proteins LRG_321p1:p.R175H LRG_321p3:p.R175H LRG_321p4:p.R175H LRG_321p5:p.R43H LRG_321p6:p.R43H LRG_321p7:p.R43H LRG_321p8:p.R136H LRG_321p9:p.R136H LRG_321p10:p.R136H LRG_321p11:p.R16H LRG_321p12:p.R16H LRG_321p12:p.R16H	TP53 alpha p.R175H TP53 beta p.R175H TP53 gamma p.R175H TP53 domain HCD III - DNA binding Structural motif Loop L2 Post-translational modifications -

Description	Comments
Records in database 3428 Classification Missense_Mutation Comment Exonic mutation Frequency This single nucleotide variant is very frequent	Activity Inactive Isoforms This substitution targets 12 TP53 isoforms Prediction Damaging

Quick Start: deletion (cDNA)

Reference is **NM_000546.5 (full Length RNA)** position is **529** to **546**
event is **Del of 18 nucleotides**

! Nomenclature of the TP53 cDNA goes forward

The screenshot shows a variant caller interface with the following fields and steps:

- 1** Reference sequence: GRCh37 (hg19)
- 2** Variant type: Deletion
- 3** Start position (larger than end position): 7578223
- 4** End position: 7578222
- 5** Wild type allele (coding strand): GA
- Mutant allele (coding strand): e.g., ATGC...
- Strand polarity (leave positive if you have no information): Positive
- Sample ID (optional): Sample ID
- 6** Start the analysis

1 Select the reference sequence

- NCBI36 (hg18)
- GRCh37 (hg19)
- GRCh38 (hg38)
- ✓ NM_000546.5 (RNA)

→ **NM_000546.5**

2 Select Deletion as a variation type

3 Enter the start position → 529

4 Enter the end position → 546

5 Deleted nucleotides are automatically displayed

6 Push Start →

Reference	Start	End	Wild type	Mutant	Type
HG18	7519131	7519131	G	A	SNP

Nomenclature	Full description according to LRG	TP53 proteins
Mutation in HGVS format NG_017013.2:g.17463G>A NCBI36 (hg18) chr17:g.7519131G>A GRCh37 (hg19) chr17:g.7578406G>A GRCh38 (hg38) chr17:g.7675088G>A	Affected transcripts LRG_321t1:c.524G>A LRG_321t2:c.524G>A LRG_321t3:c.524G>A LRG_321t4:c.524G>A LRG_321t5:c.128G>A LRG_321t6:c.128G>A LRG_321t7:c.128G>A LRG_321t8:c.407G>A Affected proteins LRG_321p1:p.R175H LRG_321p3:p.R175H LRG_321p4:p.R175H LRG_321p5:p.R43H LRG_321p6:p.R43H LRG_321p7:p.R43H LRG_321p8:p.R136H LRG_321p9:p.R136H LRG_321p10:p.R136H LRG_321p11:p.R16H LRG_321p12:p.R16H LRG_321p12:p.R16H	TP53 alpha p.R175H TP53 beta p.R175H TP53 gamma p.R175H TP53 domain HCD III - DNA binding Structural motif Loop L2 Post-translational modifications -

Description	Comments
Records in database 3428 Classification Missense_Mutation Comment Exonic mutation Frequency This single nucleotide variant is very frequent	Activity Inactive Isoforms This substitution targets 12 TP53 isoforms Prediction Damaging

Quick Start: Insertion/deletion (genomic)

Reference is **hg19**

position is **7579580** to **7579577**

event is **del4ins GTCGT**

! Nomenclature of the TP53 gene goes backward

The screenshot shows a variant caller interface with the following fields and callouts:

- 1**: Reference sequence dropdown menu.
- 2**: Variant type dropdown menu.
- 3**: Start position input field.
- 4**: End position input field.
- 5**: Mutant allele (coding strand) input field.
- 6**: Start the analysis button.

1 Select the reference sequence

NCBI36 (hg18)
 ✓ GRCh37 (hg19)
 GRCh38 (hg38)
 NM_000546.5 (RNA)

→ **hg19**

2 Select Indel as a variation type

3 Enter the start position of the deletion → 7579580

4 Enter the end position of the deletion → 7579577

5 Enter the inserted nucleotides → **GTCGT**

Check carefully the orientation of the inserted sequence as the nomenclature of the TP53 gene goes backward

6 Push Start →

Reference	Start	End	Wild type	Mutant	Type
HG19	7579580	7579577	CGTC	GTCGT	INDEL

Nomenclature

Mutation in HGVS format
 NG_017013.2:g.16289_16292delinsGTCGT

NCBI36 (hg18)
 chr17:g.7520305_7520302delinsGTCGT

GRCh37 (hg19)
 chr17:g.7579580_7579577delinsGTCGT

GRCh38 (hg38)
 chr17:g.7676262_7676259delinsGTCGT

Full description according to LRG

Affected transcripts

LRG_321t1:c.107_110delinsGTCGT
 LRG_321t2:c.107_110delinsGTCGT
 LRG_321t3:c.107_110delinsGTCGT
 LRG_321t4:c.107_110delinsGTCGT
 LRG_321t5:c.-1047_-1044delinsGTCGT
 LRG_321t6:c.-1047_-1044delinsGTCGT
 LRG_321t7:c.-1047_-1044delinsGTCGT
 LRG_321t8:c.-11_-8delinsGTCGT

Affected proteins

LRG_321p1:p.(P36Rfs*7)
 LRG_321p3:p.(P36Rfs*7)
 LRG_321p4:p.(P36Rfs*7)
 LRG_321p5:p.(=)
 LRG_321p6:p.(=)
 LRG_321p7:p.(=)
 LRG_321p8:p.(=)
 LRG_321p9:p.(=)
 LRG_321p10:p.(=)
 LRG_321p11:p.(=)
 LRG_321p12:p.(=)
 LRG_321p12:p.(=)

TP53 proteins

TP53 alpha p.(P36Rfs*7)

TP53 beta p.(P36Rfs*7)

TP53 gamma p.(P36Rfs*7)

TP53 domain Transactivation TAD1

Structural motif -

Post-translational modifications -

Description

Records in database
3

Classification
Frameshift_Indel

Comment
Exonic mutation

Frequency
 This frameshift variant is not frequent

Comments

Activity
 The consequences of this complex indel are unknown; the activity of truncated TP53 is assumed to be nil

Isoforms
 -

Prediction
 No prediction for frameshift mutation

Quick Start: Duplication (genomic)

Reference is **hg19**

position is **7579364** to **7579358**

event is **dupGTTTCCG**

! Nomenclature of the TP53 gene goes backward

Reference sequence: GRCh37 (hg19) **1**

Variant type: Duplication **2**

Start position (larger than end position): 7579364 **3**

End position: 7579358 **4**

Wild type allele (coding strand): GTTTCCG **5**

Mutant allele (coding strand): e.g., ATGC...

Strand polarity (leave positive if you have no information): Positive

Sample ID (optional): Sample ID

6 Start the analysis

1 Select the reference sequence

- NCBI36 (hg18)
- ✓ GRCh37 (hg19) → **hg19**
- GRCh38 (hg38)
- NM_000546.5 (RNA)

2 Select Duplication as a variation type

3 Enter the start position of the duplicaton → **7579364**

4 Enter the end position of the duplication → **7579358**

5 Duplicated nucleotides are automatically displayed → **GTTTCCG**

Check carefully the orientation of the duplicated sequence as the nomenclature of the TP53 gene goes backward

6 Push Start →

Reference	Start	End	Wild type	Mutant	Type
HG19	7579364	7579358	GTTTCCG	GTTTCCG	DUP

Nomenclature	Full description according to LRG	TP53 proteins
Mutation in HGVS format NG_017013.2:g.16505_16511dup NCBI36 (hg18) chr17:g.7520089_7520083dupGTTTCCG GRCh37 (hg19) chr17:g.7579364_7579358dupGTTTCCG GRCh38 (hg38) chr17:g.7676046_7676040dupGTTTCCG	Affected transcripts LRG_321t1:c.323_329dup LRG_321t2:c.323_329dup LRG_321t3:c.323_329dup LRG_321t4:c.323_329dup LRG_321t5:c.831_825dup LRG_321t6:c.831_825dup LRG_321t7:c.831_825dup LRG_321t8:c.206_212dup Affected proteins LRG_321p1:p.(L111Ffs*40) LRG_321p3:p.(L111Ffs*40) LRG_321p4:p.(L111Ffs*40) LRG_321p5:p(=) LRG_321p6:p(=) LRG_321p7:p(=) LRG_321p8:p.(L72Ffs*40) LRG_321p9:p.(L72Ffs*40) LRG_321p10:p.(L72Ffs*40) LRG_321p11:p(=) LRG_321p12:p(=) LRG_321p12:p(=)	TP53 alpha p.(L111Ffs*40) TP53 beta p.(L111Ffs*40) TP53 gamma p.(L111Ffs*40) TP53 domain DNA binding Structural motif - Post-translational modifications -

Description
Records in database 5
Classification Frame_Shift_Ins
Comment Exonic mutation
Frequency This frameshift variant is frequent

Comments
Activity The activity of truncated p53 is assumed to be nil
Isoforms -
Prediction No prediction for frameshift mutation

Quick Start: Duplication (cDNA)

Reference is **NM_000546.5 (full Length RNA)** position is **473** to **478**
event is **dupGTTTCCG**

! Nomenclature of the TP53 cDNA goes forward

Reference sequence: NM_000546.5 (RNA) 1

Variant type: Duplication 2

Start position (smaller than end position): 473 3

End position: 478 4

Wild type allele (coding strand): GCGCCA 5

Mutant allele (coding strand): e.g., ATGC...

Strand polarity (leave positive if you have no information): Positive

Sample ID (optional): Sample ID

6 Start the analysis

1 Select the reference sequence

NCBI36 (hg18)
GRCh37 (hg19)
GRCh38 (hg38)
✓ NM_000546.5 (RNA)

→ **NM_000546.5**

2 Select Duplication as a variation type

3 Enter the start position of the duplicaton

→ **473**

4 Enter the end position of the duplication

→ **478**

5 Duplicated nucleotides are automatically displayed

→ **GCGCCA**

Check carefully the orientation of the duplicated sequence as the nomenclature of the TP53 gene goes backward

6 Push Start



Reference	Start	End	Wild type	Mutant	Type
NM_000546.5	473	478	GCGCCA	GCGCCA	DUP

Nomenclature

Mutation in HGVS format
NG_017013.2:g.17412_17417dup
NCBI36 (hg18)
chr17:g.7519182_7519177dupGCGCCA
GRCh37 (hg19)
chr17:g.7578457_7578452dupGCGCCA
GRCh38 (hg38)
chr17:g.7675139_7675134dupGCGCCA

Description

Records in database
3

Classification
In_Frame_Ins

Comment
Exonic mutation

Frequency
This frameshift variant is not frequent

Full description according to LRG

Affected transcripts
LRG_32111:c.473_478dup
LRG_32112:c.473_478dup
LRG_32113:c.473_478dup
LRG_32114:c.473_478dup
LRG_32115:c.77_82dup
LRG_32116:c.77_82dup
LRG_32117:c.77_82dup
LRG_32118:c.356_361dup

Affected proteins
LRG_321p1:p.(A159_M160insSA)
LRG_321p3:p.(A159_M160insSA)
LRG_321p4:p.(A159_M160insSA)
LRG_321p5:p.(A27_M28insSA)
LRG_321p6:p.(A27_M28insSA)
LRG_321p7:p.(A27_M28insSA)
LRG_321p8:p.(A120_M121insSA)
LRG_321p9:p.(A120_M121insSA)
LRG_321p10:p.(A120_M121insSA)
LRG_321p11:p.(M17)
LRG_321p12:p.(M17)
LRG_321p12:p.(M17)

TP53 proteins

TP53 alpha
p.(A159_M160insSA)

TP53 beta
p.(A159_M160insSA)

TP53 gamma
p.(A159_M160insSA)

TP53 domain
DNA binding

Structural motif
Beta Strand S4

Post-translational modifications
-

Comments

Activity
The consequences of this in-frame insertion are unknown

Isoforms
-

Prediction
No prediction for frameshift mutation

Quick Start: Insertion/deletion (cDNA)

Reference is **NM_000546.5** (full Length RNA) position is **741** to **742**
event is **delCCinsTT**

! Nomenclature of the TP53 cDNA goes forward

The screenshot shows a variant entry form with the following fields and values:

- Reference sequence: NM_000546.5 (RNA) (1)
- Variant type: Indel (insertion/deletion) (2)
- Start position: 741 (3)
- End position: 742 (4)
- Wild type allele (coding strand): CC
- Mutant allele (coding strand): TT (5)
- Strand polarity: Positive
- Sample ID: (empty)
- Start the analysis button (6)

1 Select the reference sequence

NCBI36 (hg18)
GRCh37 (hg19)
GRCh38 (hg38)
✓ NM_000546.5 (RNA)

→ **NM_000546.5**

2 Select Indel as a variation type

3 Enter the start position of the deletion → **741**

4 Enter the end position of the deletion → **742**

5 Enter the inserted nucleotides → **TT**

Check carefully the orientation of the inserted sequence

6 Push Start →

The screenshot shows the analysis results for the variant NM_000546.5:c.741_742delinsTT. The results are organized into several panels:

- Input data:** Reference: NM_000546.5, Start: 741, End: 742, Wild type: CC, Mutant: TT, Type: INDEL.
- Nomenclature:** Mutation in HGVS format: NG_017013.2:g.18329_18330delinsTT. Other identifiers include NCBI36 (hg18), GRCh37 (hg19), and GRCh38 (hg38).
- Description:** Records in database: 43. Classification: Missense_Mutation. Comment: Exonic mutation. Frequency: This frameshift variant is very frequent.
- Full description according to LRG:** Lists affected transcripts (LRG_32111 to LRG_32118) and affected proteins (LRG_321p1 to LRG_321p12).
- TP53 proteins:** Lists TP53 alphas, betas, and gamma, along with domain information (HCD IV - DNA binding) and structural motifs (Loop L3).
- Comments:** Activity: No activity. Isoforms: -. Prediction: No prediction for frameshift mutation.

Quick Start: Insertion (genomic)

Reference is **hg19**

position is **7578223** to **7578222**

event is **Ins of 1 nucleotide (G)**



Nomenclature of the TP53 gene goes backward

The screenshot shows a variant caller interface with the following fields and callouts:

- 1**: Reference sequence dropdown menu showing "GRCh37 (hg19)".
- 2**: Variant type dropdown menu showing "Insertion".
- 3**: Start position input field containing "7579585".
- 4**: Wild type allele (coding strand) dropdown menu showing "CT".
- 5**: Mutant allele (coding strand) input field containing "G".
- 6**: "Start the analysis" button.

1 Select the reference sequence

NCBI36 (hg18)
✓ GRCh37 (hg19) → **hg19**
GRCh38 (hg38)
NM_000546.5 (RNA)

2 Select Insertion as a variation type

3 Enter the start position → **7579585**

4 End position is automatically displayed → **7579584**

5 Enter the inserted nucleotides → **G**

Check carefully the orientation of the inserted sequence as the nomenclature of the TP53 gene goes backward

6 Push Start →

The screenshot shows the variant analysis results page with the following sections:

- Input data**: Reference (HG19), Start (7579585), End (7579584), Wild type (CT), Mutant (G), Type (INS).
- Nomenclature**: Mutation in HGVS format: NG_017013.2:g.16284_16285insG; NCBI36 (hg18): chr17:g.7520310_7520309insG; GRCh37 (hg19): chr17:g.7579585_7579584insG; GRCh38 (hg38): chr17:g.7676267_7676266insG.
- Description**: Records in database: 4; Classification: Frame_Shift_Ins; Comment: Exonic mutation; Frequency: This frameshift variant is not frequent.
- Full description according to LRG**: Affected transcripts: LRG_321t1:c.102_103insG, LRG_321t2:c.102_103insG, LRG_321t3:c.102_103insG, LRG_321t4:c.102_103insG, LRG_321t5:c.-1052_-1051insG, LRG_321t6:c.-1052_-1051insG, LRG_321t7:c.-1052_-1051insG, LRG_321t8:c.-16_-15insG; Affected proteins: LRG_321p1:p.(L35Vfs*8), LRG_321p3:p.(L35Vfs*8), LRG_321p4:p.(L35Vfs*8), LRG_321p5:p(=), LRG_321p6:p(=), LRG_321p7:p(=), LRG_321p8:p(=), LRG_321p9:p(=), LRG_321p10:p(=), LRG_321p11:p(=), LRG_321p12:p(=).
- TP53 proteins**: TP53 alpha p.(L35Vfs*8), TP53 beta p.(L35Vfs*8), TP53 gamma p.(L35Vfs*8), TP53 domain Transactivation TAD1, Structural motif, Post-translational modifications.
- Comments**: Activity: The activity of truncated p53 is assumed to be nil; Isoforms: -; Prediction: No prediction for frameshift mutation.

Quick Start: Insertion (cDNA)

Reference is **NM_000546.5 (full Length RNA)** position is **328 to 329**
event is **Ins of 5 nucleotides (AAACC)**

! Nomenclature of the TP53 cDNA goes forward

The screenshot shows a variant entry form with the following fields and steps:

- 1** Reference sequence: NM_000546.5 (RNA)
- 2** Variant type: Insertion
- 3** Start position (smaller than end position): 328
- 4** Wild type allele (coding strand): CG
- 5** Mutant allele (coding strand): AAACC
- 6** Start the analysis

1 Select the reference sequence

NCBI36 (hg18)
GRCh37 (hg19)
GRCh38 (hg38)
✓ NM_000546.5 (RNA)

→ **NM_000546.5**

2 Select Insertion as a variation type

3 Enter the start position → **328**

4 End position is automatically displayed → **329**

5 Enter the inserted nucleotides → **AAACC**

Check carefully the orientation of the inserted sequence

6 Push Start →

Input data					
Reference	Start	End	Wild type	Mutant	Type
NM_000546.5	328	329	CG	AAACC	INS

Nomenclature Mutation in HGVS format NG_017013.2:g.16510_16511insAAACC NCBI36 (hg18) chr17:g.7520084_7520083insAAACC GRCh37 (hg19) chr17:g.7579359_7579358insAAACC GRCh38 (hg38) chr17:g.7676041_7676040insAAACC	Full description according to LRG Affected transcripts LRG_32111:c.328_329insAAACC LRG_32112:c.328_329insAAACC LRG_32113:c.328_329insAAACC LRG_32114:c.328_329insAAACC LRG_32115:c.-826_-825insAAACC LRG_32116:c.-826_-825insAAACC LRG_32117:c.-826_-825insAAACC LRG_32118:c.211_212insAAACC Affected proteins LRG_321p1:p.(R110Qfs*15) LRG_321p3:p.(R110Qfs*15) LRG_321p4:p.(R110Qfs*15) LRG_321p5:p.(=) LRG_321p6:p.(=) LRG_321p7:p.(=) LRG_321p8:p.(R71Qfs*15) LRG_321p9:p.(R71Qfs*15) LRG_321p10:p.(R71Qfs*15) LRG_321p11:p.(=) LRG_321p12:p.(=) LRG_321p12:p.(=)	TP53 proteins TP53 alpha p.(R110Qfs*15) TP53 beta p.(R110Qfs*15) TP53 gamma p.(R110Qfs*15) TP53 domain DNA binding Structural motif Beta Strand S1 Post-translational modifications - Comments Activity The activity of truncated p53 is assumed to be nil Isoforms - Prediction No prediction for frameshift mutation
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