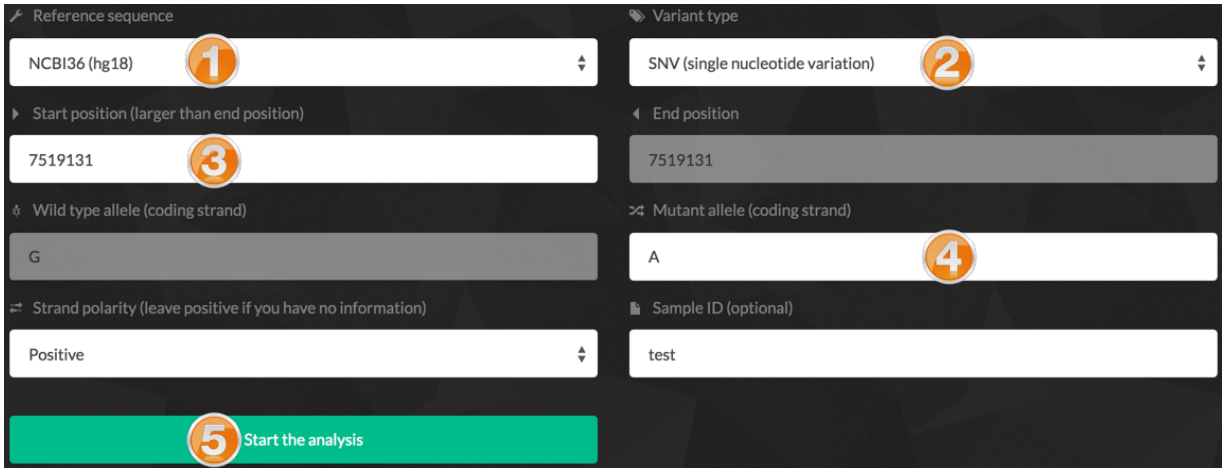
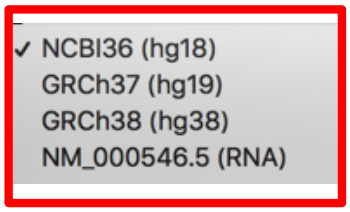
Quick Start: SNV (genomic)

Reference is hg18 position is 7518131
event is SNV mutant allele is A



1 **Select the reference sequence**



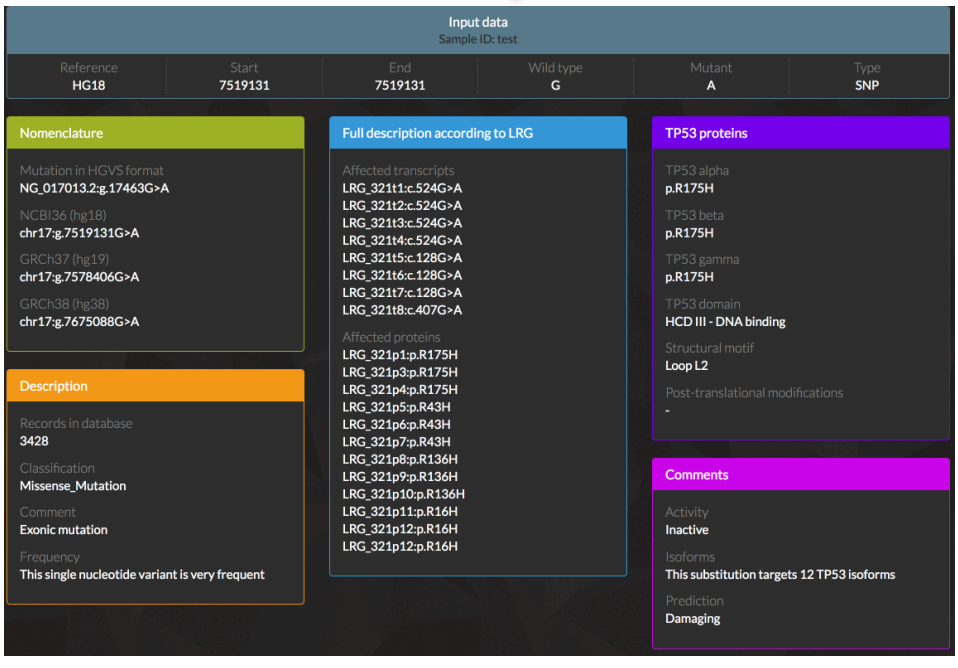
→ **hg18**

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

3 **Enter the position** → **7519131**

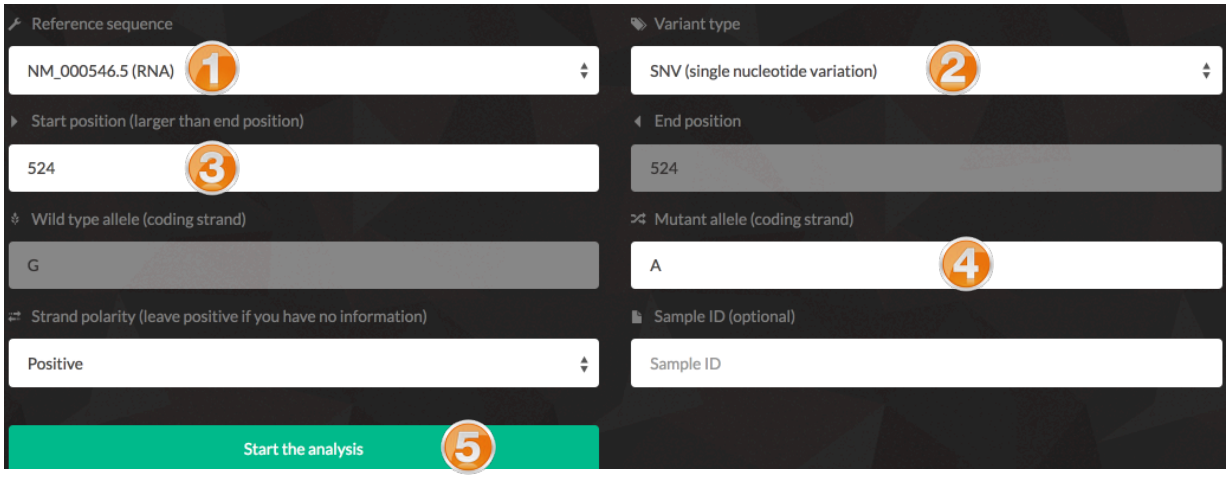
4 **Enter the mutant allele** → **A**

5 **Push Start**

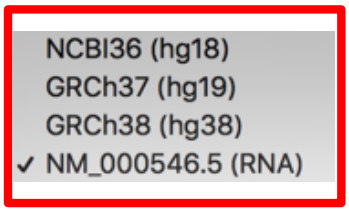


Quick Start: SNV (cDNA)

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



1 **Select the reference sequence**



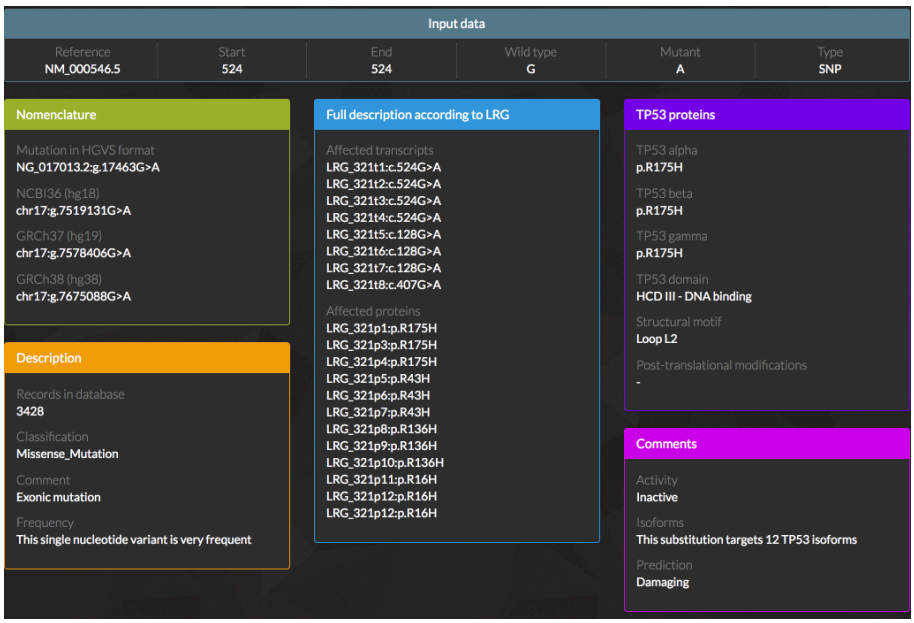
→ **NM_000546.5**

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

3 **Enter the position** → **524**

4 **Enter the mutant allele** → **A**

5 **Push Start**

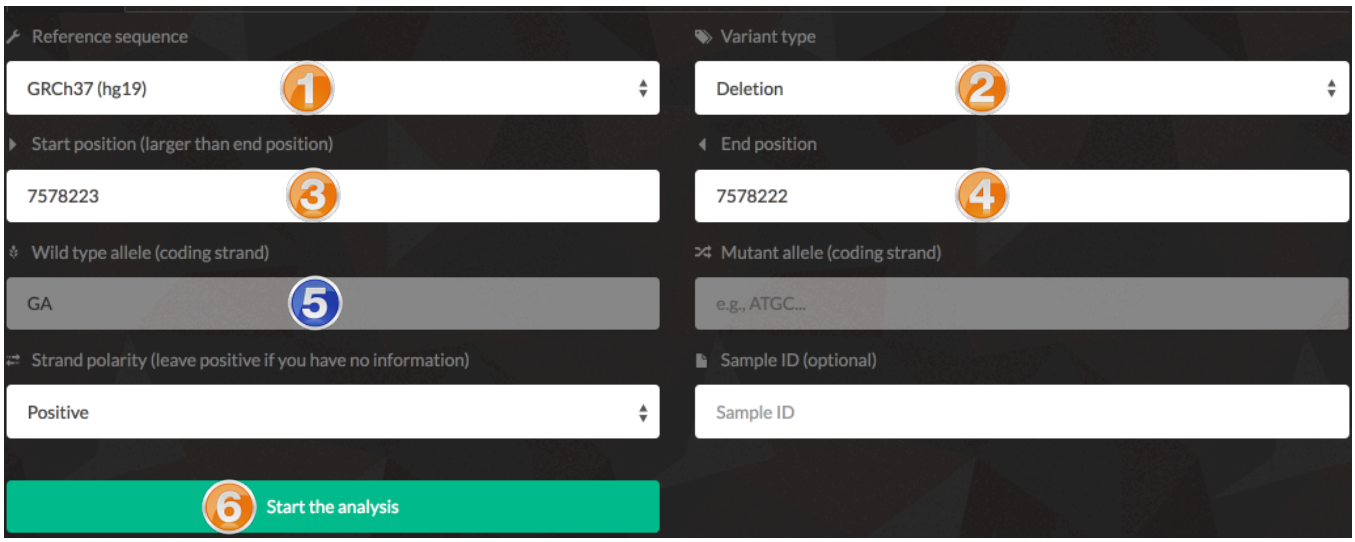


Quick Start: deletion (genomic)

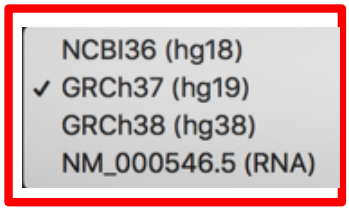
Reference is hg19 position is **7578223** to **7578222**

event is Del of 2 nucleotides

Nomenclature of the TP53 gene goes backward



1 **Select the reference sequence**



→ **hg19**

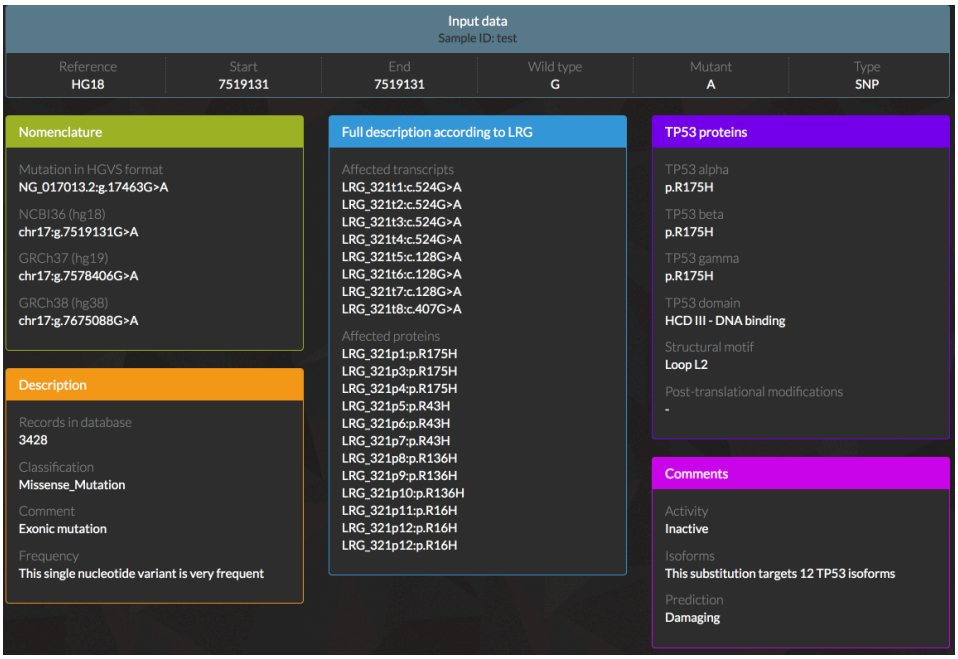
2 **Select Deletion as a variation type**

3 **Enter the start position of the deletion** → **7578223**

4 **Enter the end position of the deletion** → **7578222**

5 **Deleted nucleotides are automatically displayed**

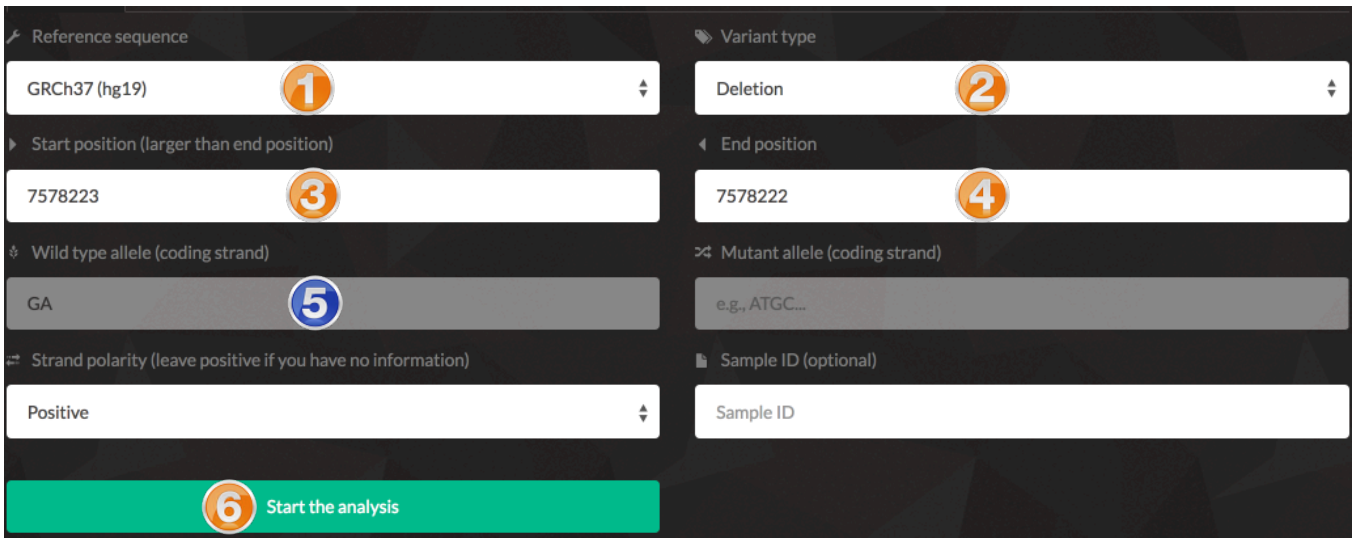
6 **Push Start** →



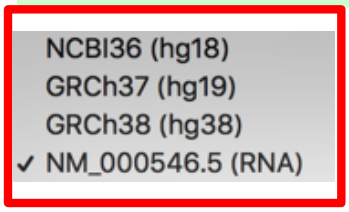
Quick Start: deletion (cDNA)

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



1 **Select the reference sequence**



→ **NM_000546.5**

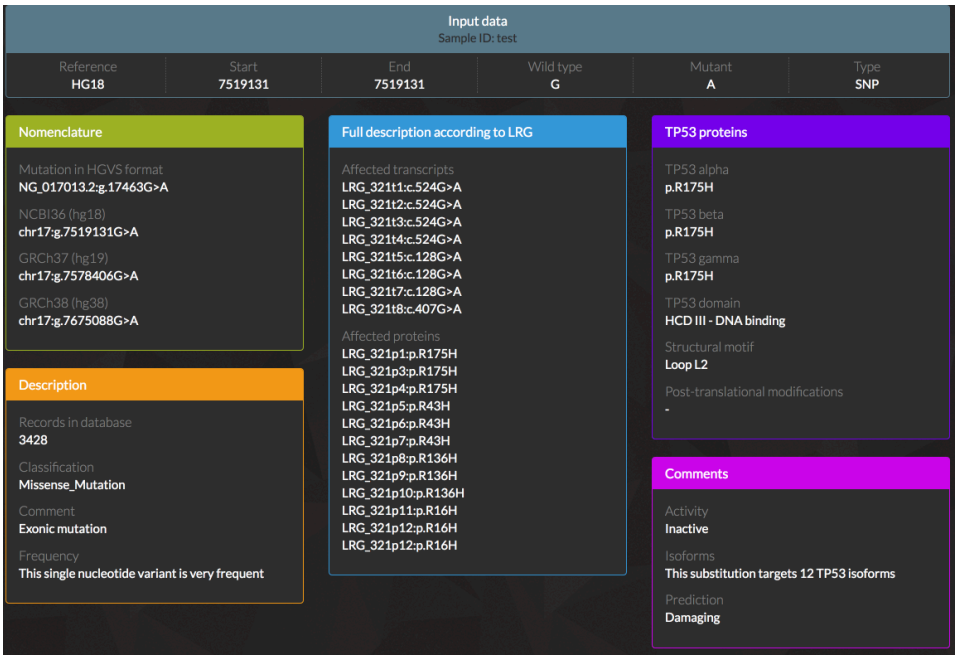
2 **Select Deletion as a variation type**

3 **Enter the start position** → **529**

4 **Enter the end position** → **546**

5 **Deleted nucleotides are automatically displayed**

6 **Push Start** →

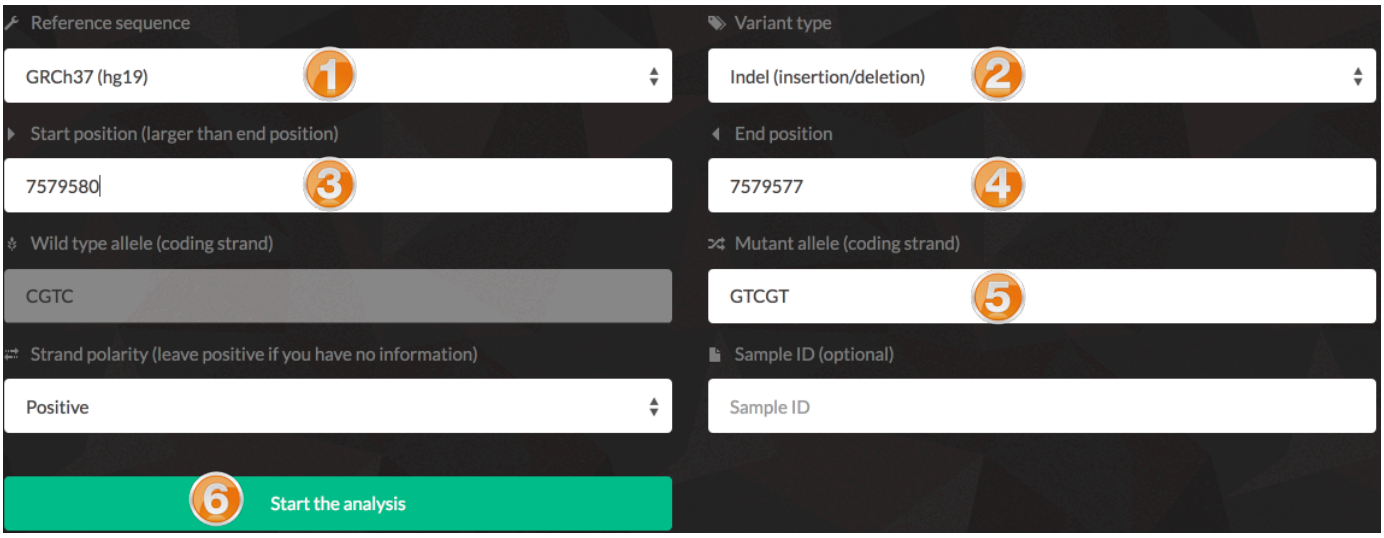


Quick Start: Insertion/deletion (genomic)

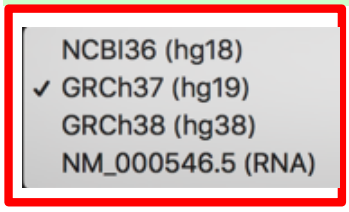
Reference is hg19 position is **7579580** to **7579577**

event is del4ins GTCGT

Nomenclature of the TP53 gene goes backward



1 **Select the reference sequence**



→ **hg19**

2 **Select Indel as a variation type**

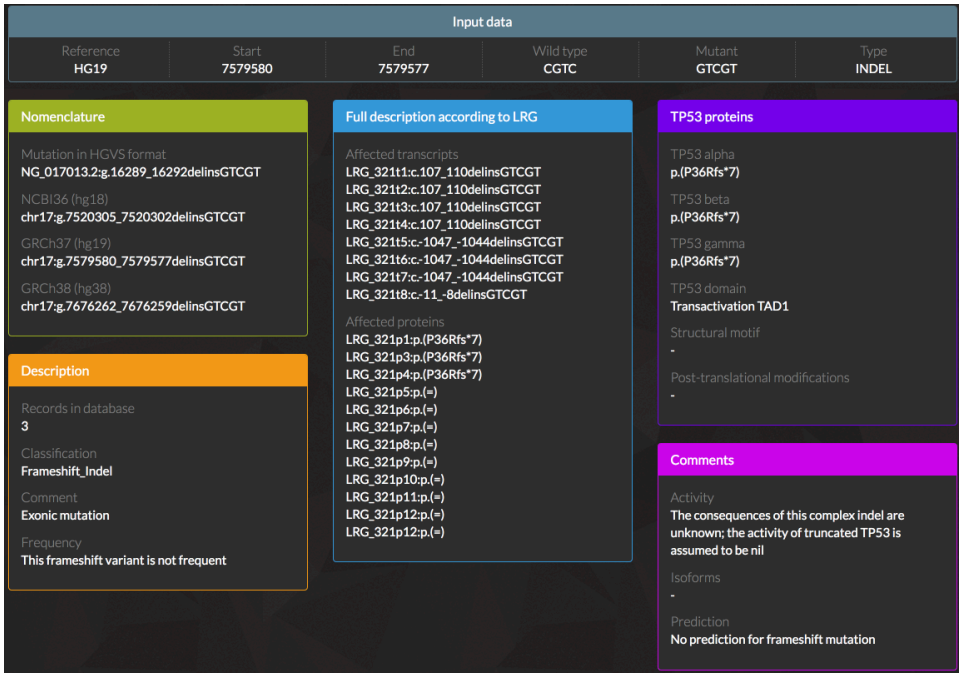
3 **Enter the start position of the deletion** → **7579580**

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** →

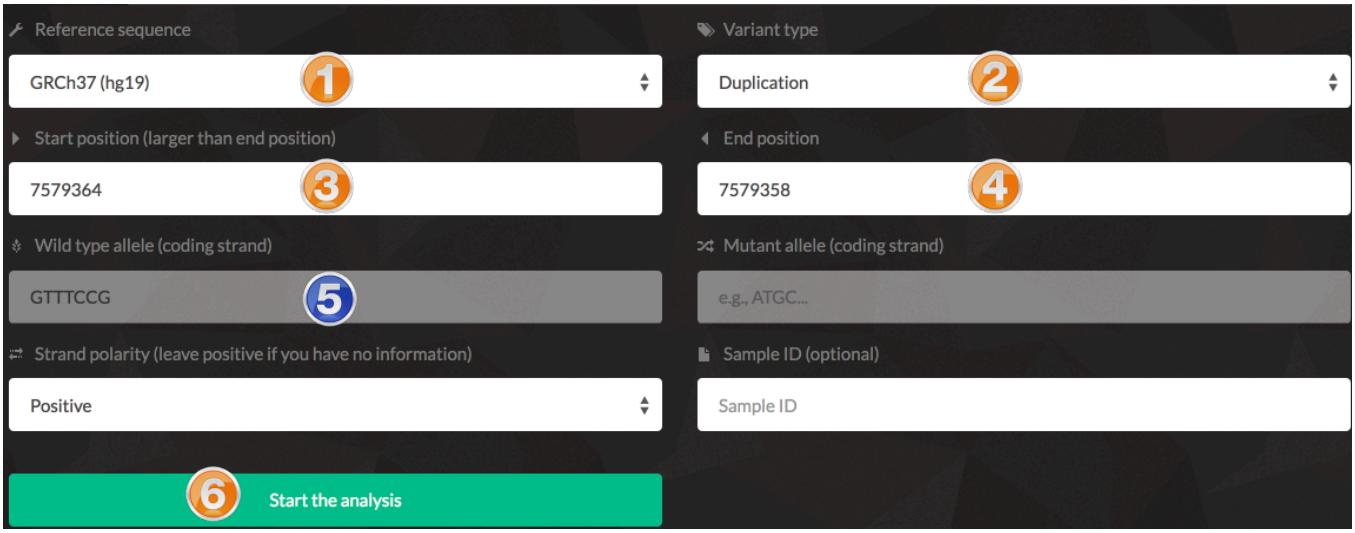


Quick Start: Duplication (genomic)

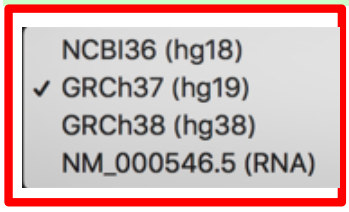
Reference is hg19 position is **7579364** to **7579358**

event is dupGTTTCCG

Nomenclature of the TP53 gene goes backward



1 **Select the reference sequence**



→ **hg19**

2 **Select Duplication as a variation type**

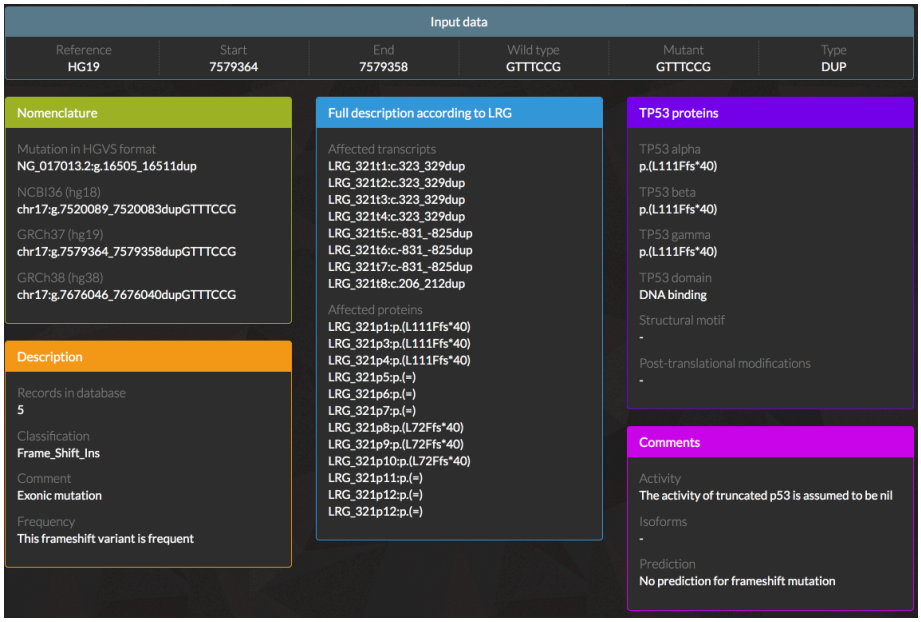
3 **Enter the start position of the duplicaton** → **7579364**

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

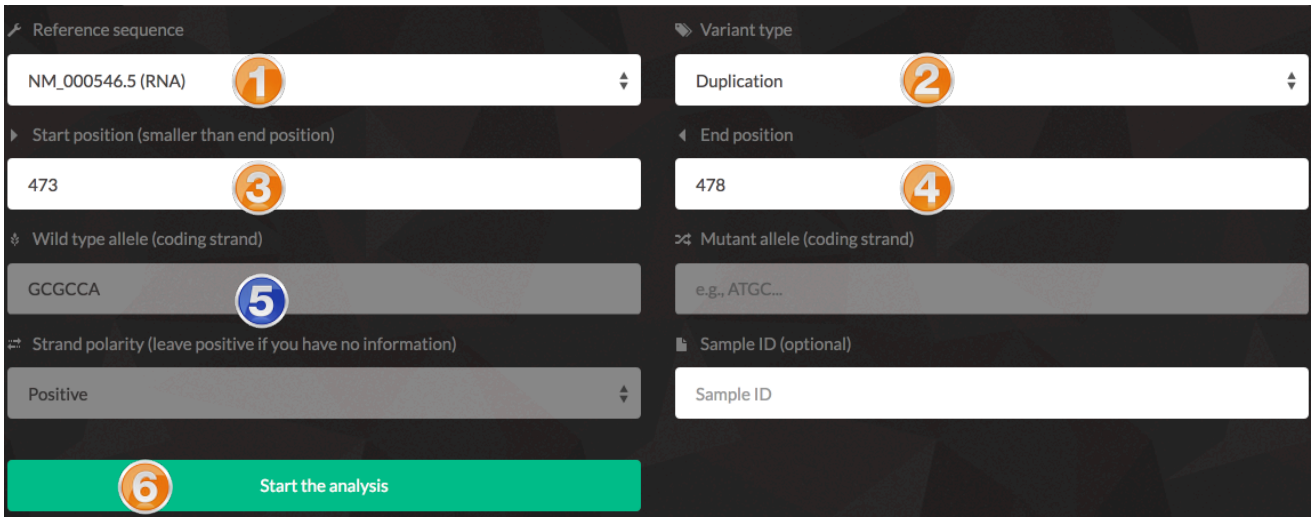
6 **Push Start** →



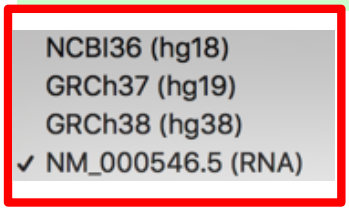
Quick Start: Duplication (cDNA)

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

Nomenclature of the TP53 cDNA goes forward



1 **Select the reference sequence**



→ **NM_000546.5**

2 **Select Duplication as a variation type**

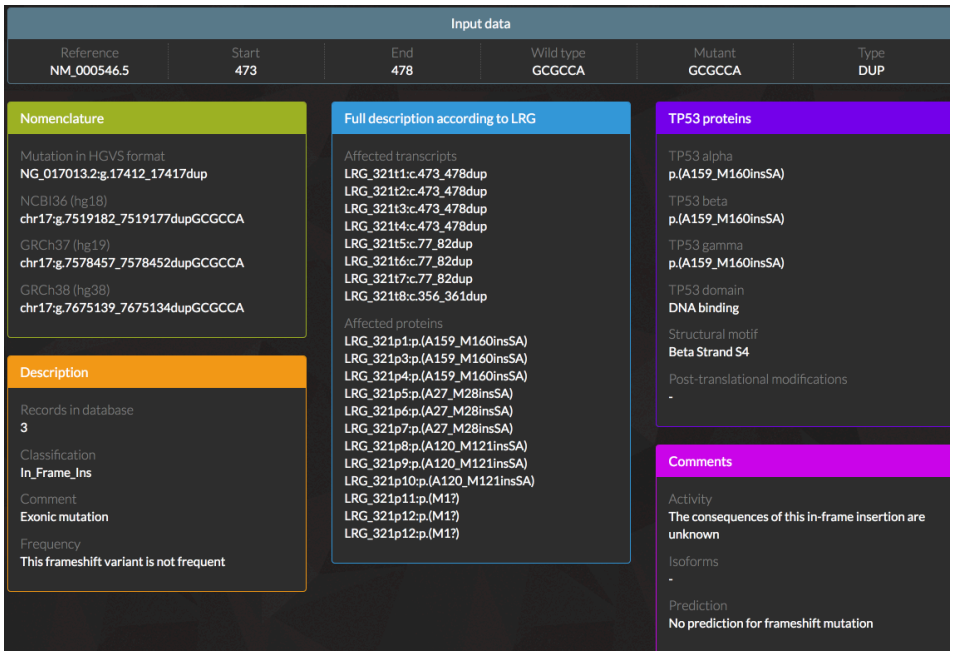
3 **Enter the start position of the duplicaton** → **473**

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

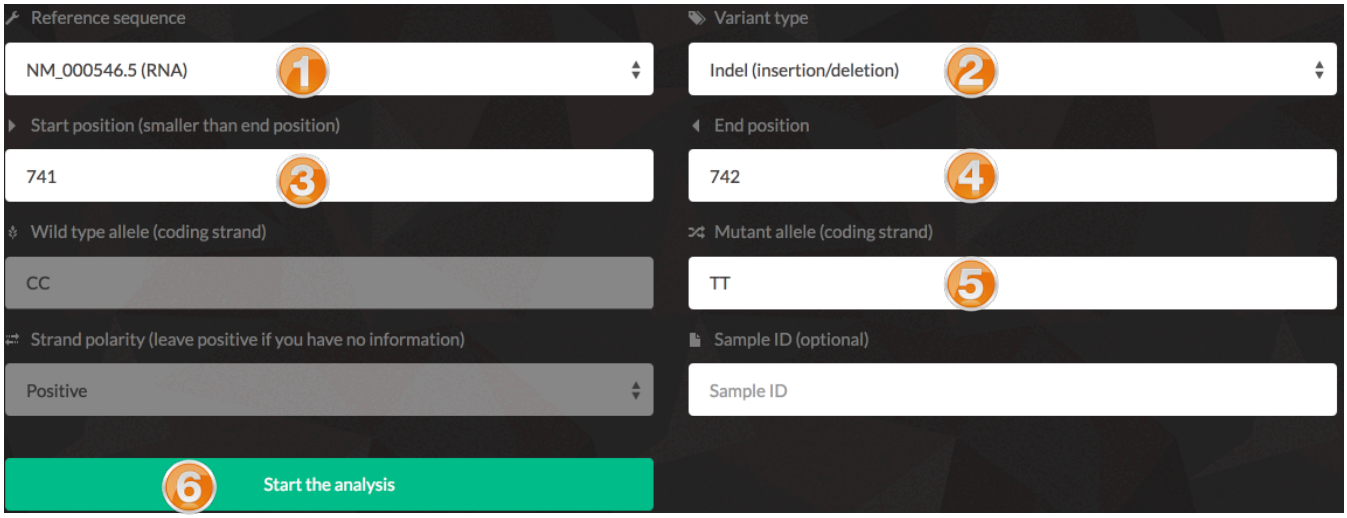
6 **Push Start** →



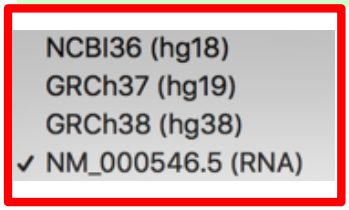
Quick Start: Insertion/deletion (cDNA)

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

Nomenclature of the TP53 cDNA goes forward



1 **Select the reference sequence**



→ **NM_000546.5**

2 **Select Indel as a variation type**

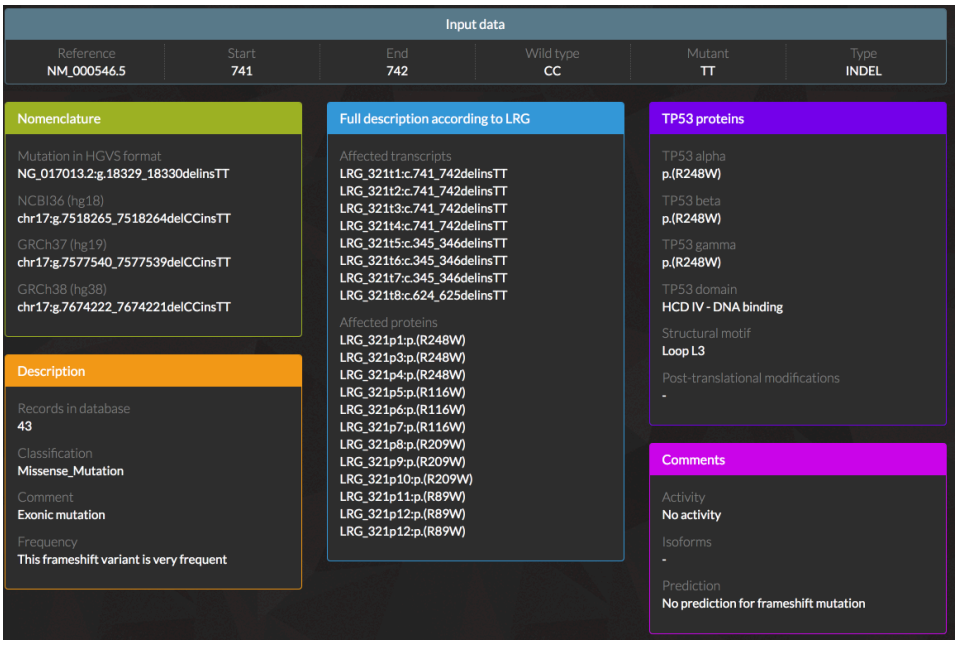
3 **Enter the start position of the deletion** → **741**

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

5 **Enter the inserted nucleotides** → **TT**

Check carefully the orientation of the inserted sequence

6 **Push Start** →

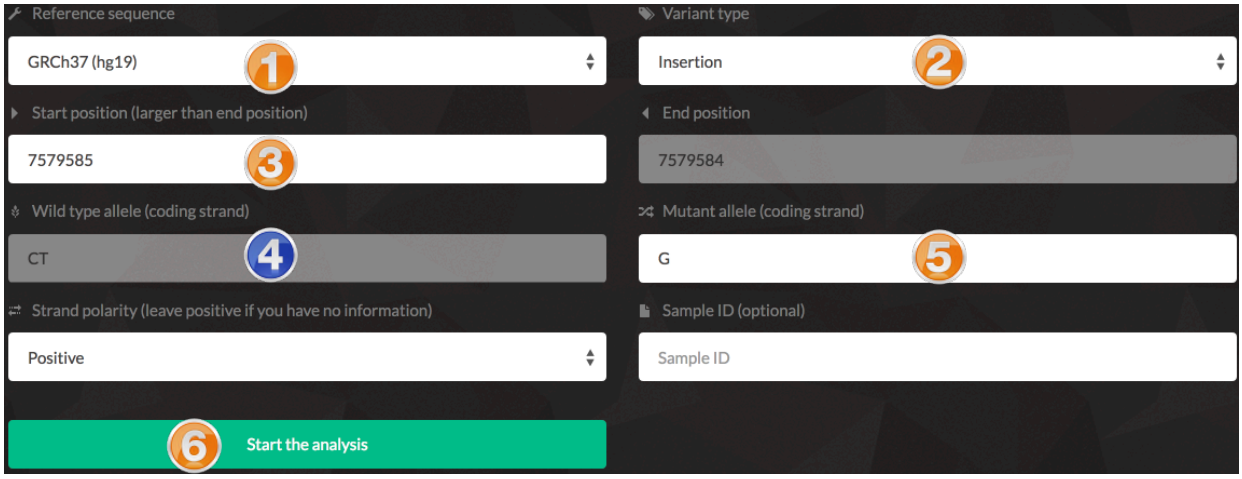


Quick Start: Insertion (genomic)

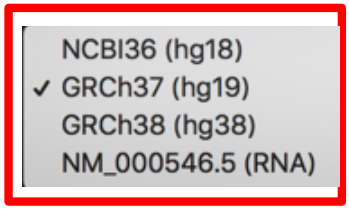
Reference is hg19 position is **7578223** to **7578222**

event is Ins of 1 nucleotide (G)

Nomenclature of the TP53 gene goes backward



Select the reference sequence



→ **hg19**

Select Insertion as a variation type

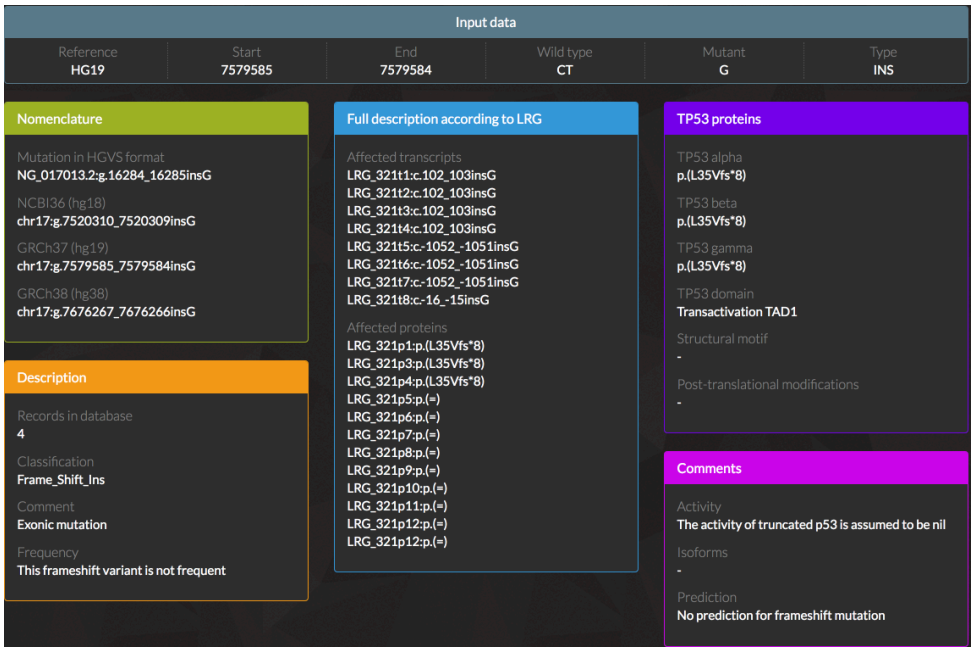
Enter the start position → **7579585**

End position is automatically displayed → **7579584**

Enter the inserted nucleotides → **G**

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

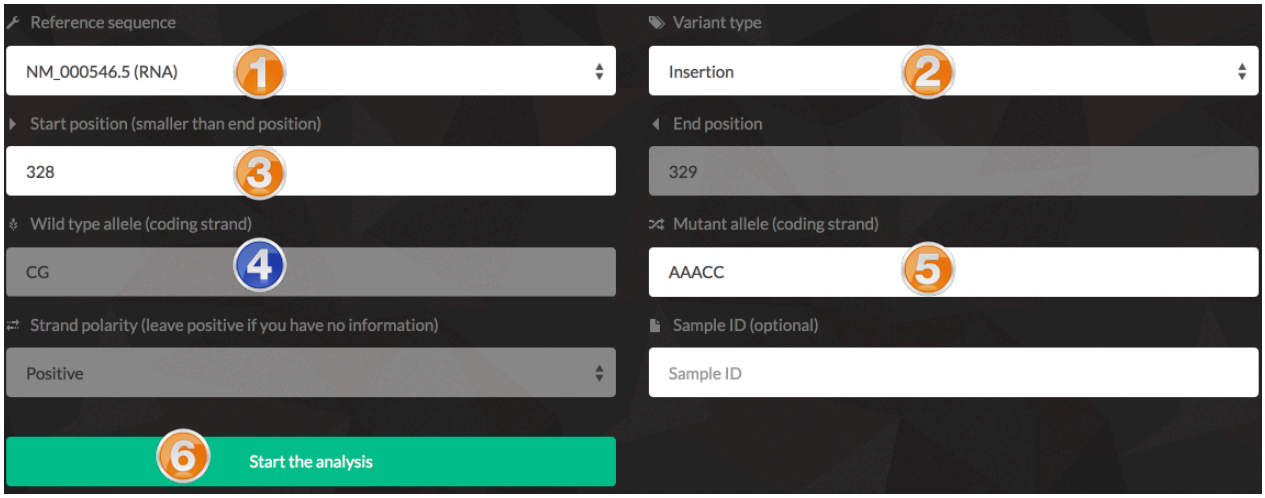
Push Start →



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



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** →