

Genetic Tests

Record ID

Genetic Tests

Genetic Test

- WES
 - Gene Panel
 - CGH
 - Single Gene
 - Karyotype
 - FISH
 - Methylation Study
 - WGS
 - Other
- (select all that apply)

WES Results

(coordinates)

Gene Panel Results

(coordinates)

CGH Lab

(coordinates)

CGH Date of Test

(coordinates)

CGH Results

(coordinates)

Single Gene Results

(coordinates)

Karyotype Lab

(coordinates)

Karyotype Date of Test

(coordinates)

Karyotype Results

(coordinates)

FISH Lab

(coordinates)

FISH Date of Test

(coordinates)

FISH Results

(coordinates)

Methylation Results

(coordinates)

WGS Results

(coordinates)

If other, what was the name of the study?

If other, what was the results of the study?

Pathologic Variants

How many pathogenic variants do you have to enter?

- 0
 1
 2
 3

Pathogenic variant 1: gene name

Pathogenic variant 1: Transcript ID

Pathogenic variant 1: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo

What is the X-linked inheritance?

- de novo
 apparent de novo
-

Pathogenic variant 1: Nucleotide Change (c.)

Pathogenic variant 1: AA change (p.)

Pathogenic variant 2: gene name

Pathogenic variant 2: Transcript ID

Pathogenic variant 2: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria
-

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo
-

What is the X-linked inheritance?

- de novo
 apparent de novo
-

Pathogenic variant 2: Nucleotide Change (c.)

Pathogenic variant 2: AA change (p.)

Pathogenic variant 3: gene name

Pathogenic variant 3: Transcript ID

Pathogenic variant 3: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria
-

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo
-

What is the X-linked inheritance?

- de novo
 apparent de novo
-

Pathogenic variant 3: Nucleotide Change (c.)

Pathogenic variant 3: AA change (p.)

Variants of Unknown Significance and 'Likely Pathogenic'

How many variants of unknown significance do you have to enter? Include 'likely pathogenic'

0
 1
 2
 3
 4
 5
 6
 7
 8
 9
 10

VOUS 1: gene name

VOUS 1: Transcript ID

VOUS 1: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo

What is the X-linked inheritance?

- de novo
 apparent de novo

VOUS 1: Nucleotide Change (c.)

VOUS 1: AA change (p.)

VOUS 2: gene name

VOUS 2: Transcript ID

VOUS 2: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo

What is the X-linked inheritance?

- de novo
 apparent de novo

VOUS 2: Nucleotide Change (c.)

VOUS 2: AA change (p.)

VOUS 3: gene name

VOUS 3: Transcript ID

VOUS 3: Inheritance pattern

- Autosomal Dominant
- Autosomal Recessive
- X-Linked
- Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
- de novo
- apparent de novo

What is the X-linked inheritance?

- de novo
- apparent de novo

VOUS 3: Nucleotide Change (c.)

VOUS 3: AA change (p.)

VOUS 4: gene name

VOUS 4: Transcript ID

VOUS 4: Inheritance pattern

- Autosomal Dominant
- Autosomal Recessive
- X-Linked
- Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
- de novo
- apparent de novo

What is the X-linked inheritance?

- de novo
- apparent de novo

VOUS 4: Nucleotide Change (c.)

VOUS 4: AA change (p.)

VOUS 5: gene name

VOUS 5: Transcript ID

VOUS 5: Inheritance pattern

- Autosomal Dominant
- Autosomal Recessive
- X-Linked
- Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
- de novo
- apparent de novo

What is the X-linked inheritance?

- de novo
- apparent de novo

VOUS 5: Nucleotide Change (c.)

VOUS 5: AA change (p.)

VOUS 6: gene name

VOUS 6: Transcript ID

VOUS 6: Inheritance pattern

- Autosomal Dominant
- Autosomal Recessive
- X-Linked
- Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
- de novo
- apparent de novo

What is the X-linked inheritance?

- de novo
- apparent de novo

VOUS 6: Nucleotide Change (c.)

VOUS 6: AA change (p.)

VOUS 7: gene name

VOUS 7: Transcript ID

VOUS 7: Inheritance pattern

- Autosomal Dominant
- Autosomal Recessive
- X-Linked
- Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
- de novo
- apparent de novo

What is the X-linked inheritance?

- de novo
 apparent de novo
-

VOUS 7: Nucleotide Change (c.)

VOUS 7: AA change (p.)

VOUS 8: gene name

VOUS 8: Transcript ID

VOUS 8: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria
-

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo
-

What is the X-linked inheritance?

- de novo
 apparent de novo
-

VOUS 8: Nucleotide Change (c.)

VOUS 8: AA change (p.)

VOUS 9: gene name

VOUS 9: Transcript ID

VOUS 9: Inheritance pattern

- Autosomal Dominant
 Autosomal Recessive
 X-Linked
 Mitochondria
-

What type of Autosomal Dominant inheritance?

- Inherited
 de novo
 apparent de novo
-

What is the X-linked inheritance?

- de novo
 apparent de novo
-

VOUS 9: Nucleotide Change (c.)

VOUS 9: AA change (p.)

VOUS 10: gene name

VOUS 10: Transcript ID

VOUS 10: Inheritance pattern

- Autosomal Dominant
- Autosomal Recessive
- X-Linked
- Mitochondria

What type of Autosomal Dominant inheritance?

- Inherited
- de novo
- apparent de novo

What is the X-linked inheritance?

- de novo
- apparent de novo

VOUS 10: Nucleotide Change (c.)

VOUS 10: AA change (p.)
