

# Mutation

## COSM4670691

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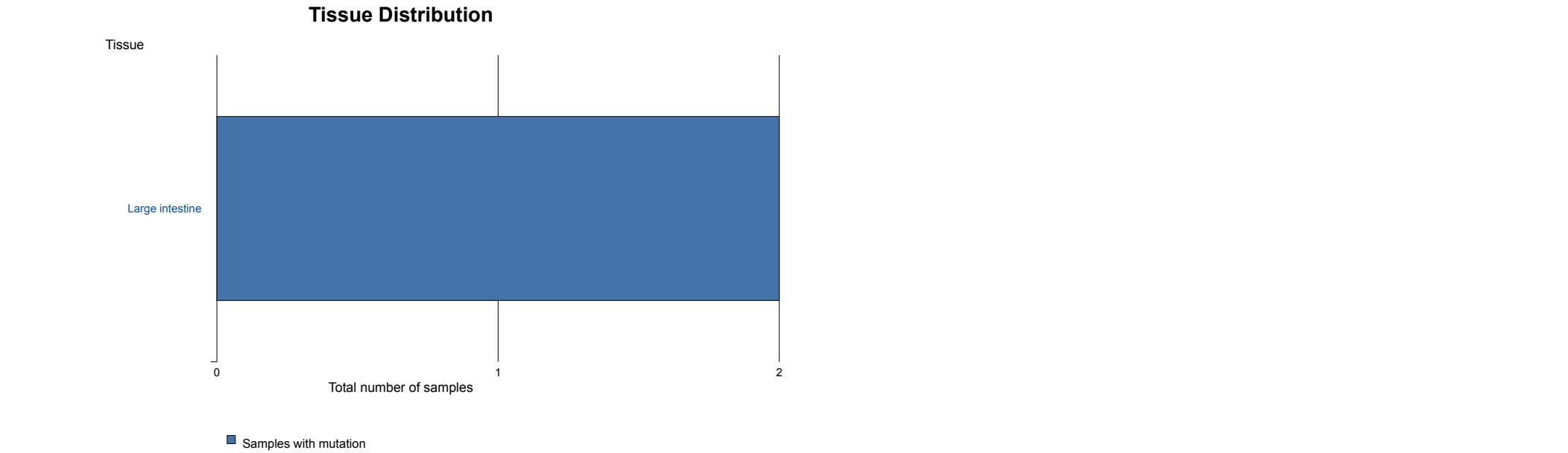
## Overview

This section shows a general overview of the selected mutation. It describes the source of the mutation i.e gene name/sample name/tissue name with unique ID, and also shows the mutation syntax at the amino acid and nucleotide sequence level. You can see more information on our [help pages](#).

Mutation ID	COSM4670691
Gene name	<a href="#">CDC42BPA_ENST00000366769</a>
AA mutation	p.R598H (Substitution - Missense, position 598, R→H)
CDS mutation	c.1793G>A (Substitution, position 1793, G→A)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">1:227112768..227112768</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

## Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



### Samples

This section displays a table of mutated samples, with tissue, histology and zygoty information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygoty	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">T464</a>	<a href="#">CDC42BPA_ENST00000366769</a>	<a href="#">ENST00000366769</a>	<a href="#">Large intestine</a>	NS	Carcinoma	Adenocarcinoma	<a href="#">27149842</a>	Unknown	Confirmed Somatic		Unknown	-	
<a href="#">T464</a>	<a href="#">CDC42BPA_ENST00000366769</a>	<a href="#">ENST00000366769</a>	<a href="#">Large intestine</a>	Colon	Carcinoma	Adenocarcinoma	<a href="#">25344691</a>	Unknown	Confirmed Somatic		Unknown	-	

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### References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
RNF43 is frequently mutated in colorectal and endometrial cancers	Giannakis M et al	2014	Nature genetics;46(12):1264-6	Curated	<a href="#">COSP37678</a>	<a href="#">25344691</a>
Genomic Correlates of Immune-Cell Infiltrates in Colorectal Carcinoma	Giannakis M et al	2016	Cell reports	Curated	<a href="#">COSP44021</a>	<a href="#">27149842</a>

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Mutation

COSM1988848

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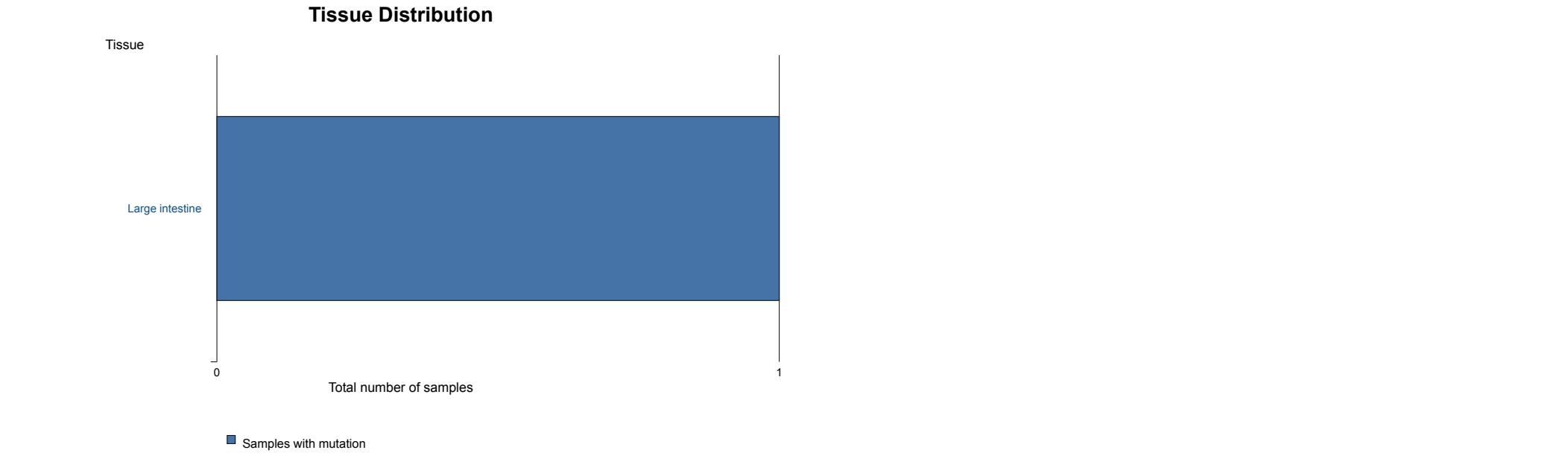
Overview

This section shows a general overview of the selected mutation. It describes the source of the mutation i.e gene name/sample name/tissue name with unique ID, and also shows the mutation syntax at the amino acid and nucleotide sequence level. You can see more information on our [help pages](#).

Mutation ID	COSM1988848
Gene name	<a href="#">R3HDM2 ENST00000347140</a>
AA mutation	p.R449H (Substitution - Missense, position 449, R→H)
CDS mutation	c.1346G>A (Substitution, position 1346, G→A)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">12:57269951..57269951</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	No
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



Samples

This section displays a table of mutated samples, with tissue, histology and zygotity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygotity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">RKO</a>	<a href="#">R3HDM2_ENST00000347140</a>	<a href="#">ENST00000347140</a>	<a href="#">Large intestine</a>	NS	Carcinoma	Adenocarcinoma	<a href="#">24755471</a>	Heterozygous	Unknown	Cultured	Unknown	-	

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References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
Colorectal cancer cell lines are representative models of the main molecular subtypes of primary cancer	Mouradov D et al	2014	Cancer research;74(12):3238-47	Curated	<a href="#">COSP35484</a>	<a href="#">24755471</a>

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COSM6699587

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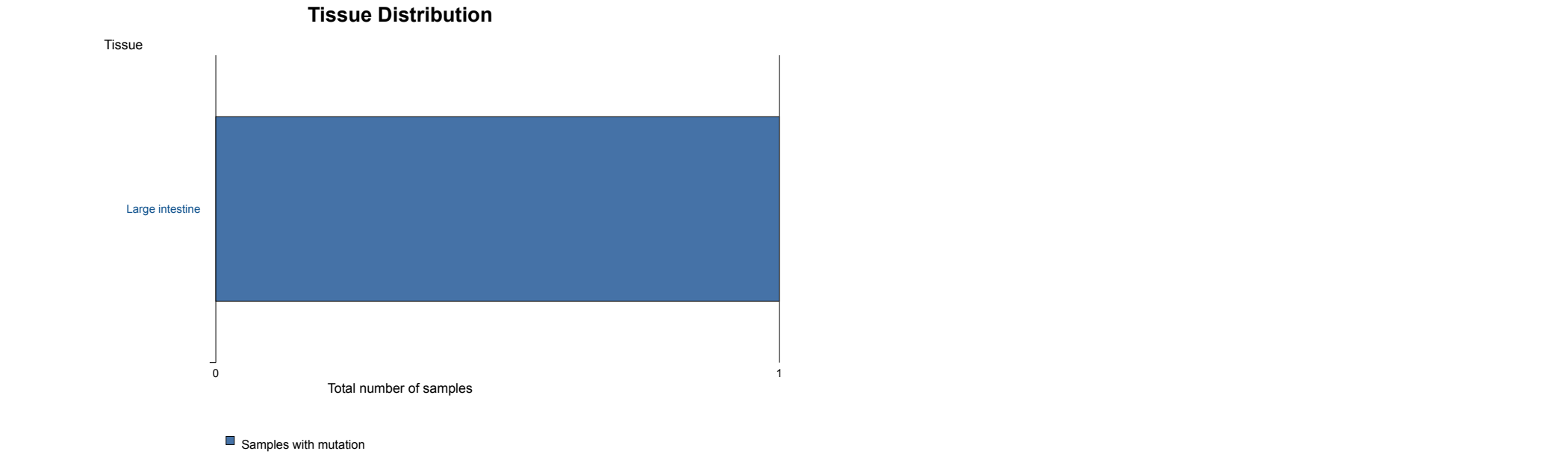
Overview

This section shows a general overview of the selected mutation. It describes the source of the mutation i.e gene name/sample name/tissue name with unique ID, and also shows the mutation syntax at the amino acid and nucleotide sequence level. You can see more information on our [help pages](#).

Mutation ID	COSM6699587
Gene name	<a href="#">LARGE ENST00000452586</a>
AA mutation	p.P435S (Substitution - Missense, position 435, P→S)
CDS mutation	c.1303C>T (Substitution, position 1303, C→T)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">22:33277227...33277227</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



### Samples

This section displays a table of mutated samples, with tissue, histology and zygosity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygosity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">T60</a>	<a href="#">LARGE_ENST00000452586</a>	<a href="#">ENST00000452586</a>	<a href="#">Large intestine</a>	NS	Carcinoma	Adenocarcinoma	<a href="#">27149842</a>	Unknown	Confirmed Somatic		Unknown	-	

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### References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
Genomic Correlates of Immune-Cell Infiltrates in Colorectal Carcinoma	Giannakis M et al	2016	Cell reports	Curated	<a href="#">COSP44021</a>	<a href="#">27149842</a>

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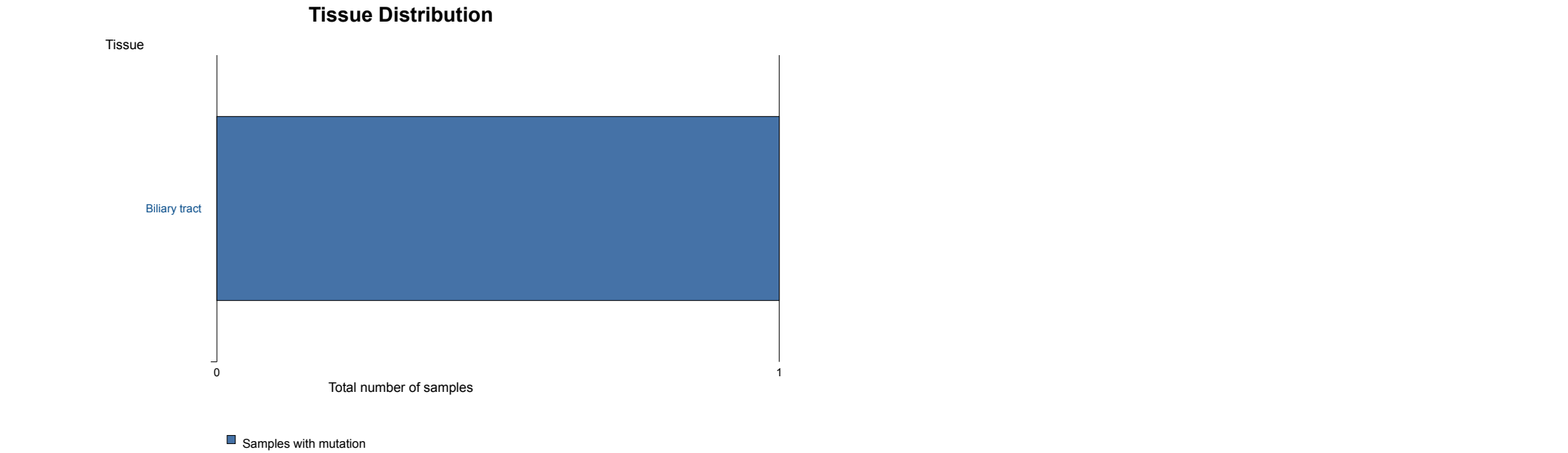
Overview

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Mutation ID	COSM5513679
Gene name	<a href="#">TOP3B_ENST00000413067</a>
AA mutation	p.? (Unknown)
CDS mutation	c.539-1G>T (Substitution - intronic)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">22:21962603..21962603</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



### Samples

This section displays a table of mutated samples, with tissue, histology and zygosity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygosity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">BD72T</a>	<a href="#">TOP3B</a>	<a href="#">ENST00000413067</a>	<a href="#">Biliary tract</a>	Bile duct	Carcinoma	NS	-	Unknown	Confirmed Somatic	Tumour Sample	Unknown	-	

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### References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
ICGC(BTCA-JP) : Biliary Tract Cancer - JP				Curated (Study)	<a href="#">COSU658</a>	

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### Mutation

### COSM4188887



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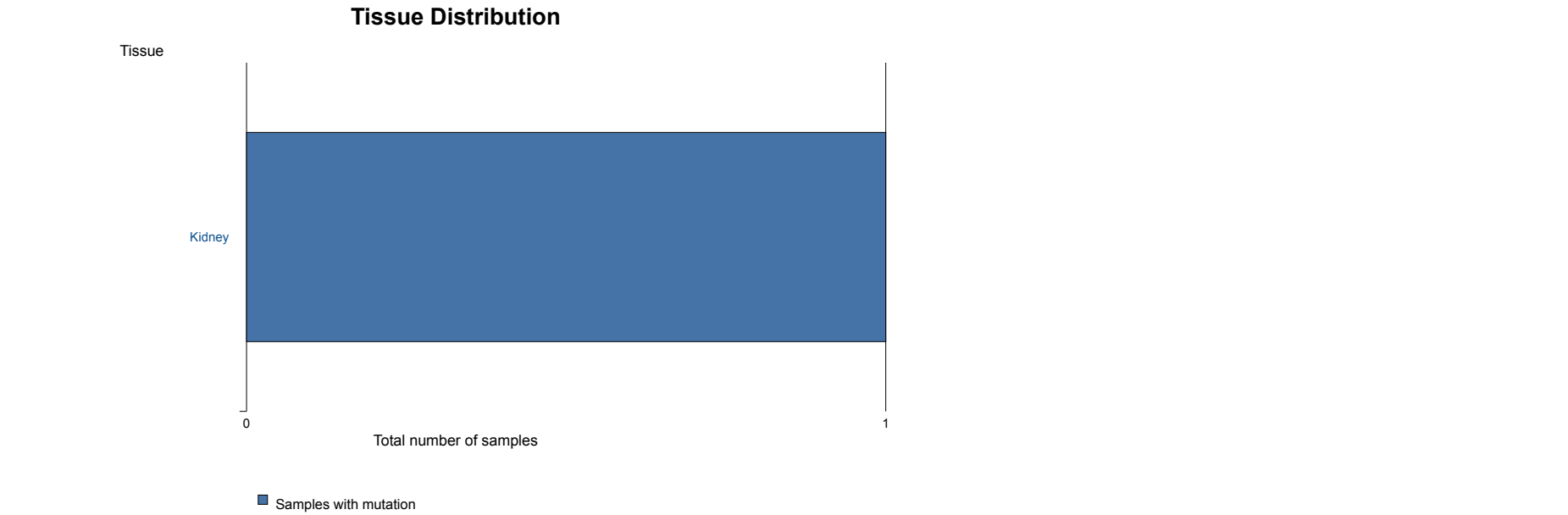
## Overview

This section shows a general overview of the selected mutation. It describes the source of the mutation i.e gene name/sample name/tissue name with unique ID, and also shows the mutation syntax at the amino acid and nucleotide sequence level. You can see more information on our [help pages](#).

Mutation ID	COSM418887
Gene name	<a href="#">HIRA</a>
AA mutation	p.T393I (Substitution - Missense, position 393, T→I)
CDS mutation	c.1178C>T (Substitution, position 1178, C→T)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">22:19385672..19385672</a> , view <a href="#">Ensembl contig</a>
CDD	<a href="#">NP_003316.3</a>
HomoloGene	<a href="#">48172</a> , view the <a href="#">multiple sequence alignment</a>
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

## Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



Samples

This section displays a table of mutated samples, with tissue, histology and zygoty information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygoty	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">BK0094</a>	<a href="#">HIRA</a>	<a href="#">ENST00000263208</a>	<a href="#">Kidney</a>	NS	Carcinoma	Clear cell renal cell carcinoma	<a href="#">24504440</a>	Unknown	Confirmed Somatic	Tumour Sample	Unknown	-	

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References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
Multilayer-omics analysis of renal cell carcinoma, including the whole exome, methylome and transcriptome	Arai E et al	2014	International journal of cancer. Journal international du cancer;135(6):1330-42	Curated	<a href="#">COSP34618</a>	<a href="#">24504440</a>

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COSM6674668

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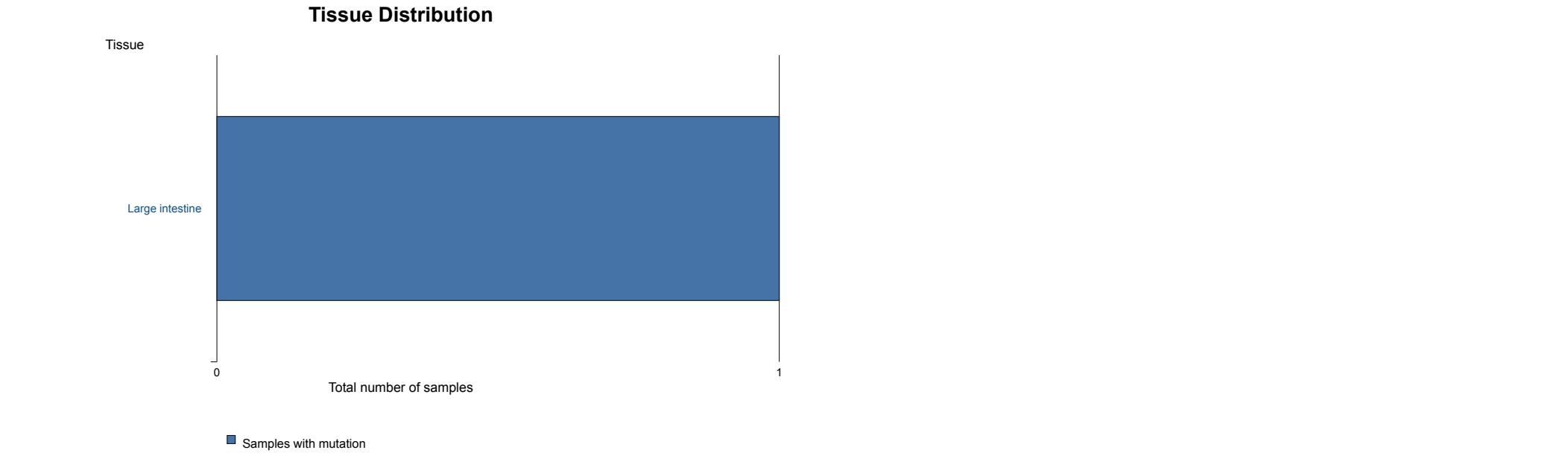
Overview

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Mutation ID	COSM6674668
Gene name	<a href="#">GPS1_ENST00000306823</a>
AA mutation	p.R449H (Substitution - Missense, position 449, R→H)
CDS mutation	c.1346G>A (Substitution, position 1346, G→A)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">17:82056919..82056919</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



Samples

This section displays a table of mutated samples, with tissue, histology and zygosity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygosity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">T237969</a>	<a href="#">GPS1_ENST00000306823</a>	<a href="#">ENST00000306823</a>	<a href="#">Large intestine</a>	NS	Carcinoma	Adenocarcinoma	<a href="#">27149842</a>	Unknown	Confirmed Somatic		Unknown	-	

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References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
Genomic Correlates of Immune-Cell Infiltrates in Colorectal Carcinoma	Giannakis M et al	2016	Cell reports	Curated	<a href="#">COSP44021</a>	<a href="#">27149842</a>

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Overview

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Mutation ID  
COSM1308066

Gene name  
[SEC14L3](#)

AA mutation  
p.E211K (Substitution - Missense, position 211, E→K)

CDS mutation  
c.631G>A (Substitution, position 631, G→A)

Nucleotides inserted  
n/a

Genomic coordinates  
GRCh38, [22:30464853..30464853](#), view [Ensembl contig](#)

CDD  
[NP\\_777635.1](#)

HomoloGene  
[24848](#), view the [multiple sequence alignment](#)

Ever confirmed somatic?  
Yes

FATHMM prediction  
Pathogenic (score 0.99)

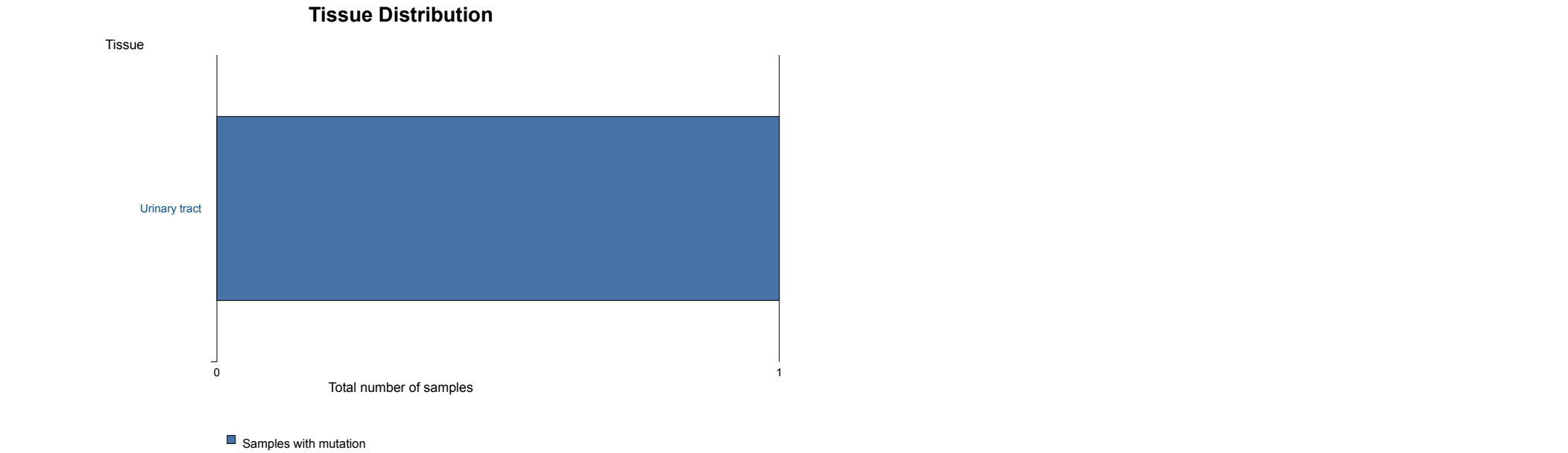
Remark  
n/a

Recurrent  
n/a

Drug resistance  
n/a

Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



Samples

This section displays a table of mutated samples, with tissue, histology and zygosity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygosity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">TCGA-DK-A3IT-01</a>	<a href="#">SEC14L3</a>	<a href="#">ENST00000215812</a>	<a href="#">Urinary tract</a>	Bladder	Carcinoma	NS	-	Unknown	Confirmed Somatic	Tumour Sample	Unknown	-	

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References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
ICGC/GDC(BLCA-US) : Bladder Urothelial Carcinoma - TCGA, US				Curated (Study)	<a href="#">COSU413</a>	

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COSM4512219

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Overview

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Mutation ID  
COSM4512219

Gene name  
[SEC14L3](#)

AA mutation  
p.P299S (Substitution - Missense, position 299, P→S)

CDS mutation  
c.895C>T (Substitution, position 895, C→T)

Nucleotides inserted  
n/a

Genomic coordinates  
GRCh38, [22:30461571..30461571](#), view [Ensembl contig](#)

CDD  
[NP\\_777635.1](#)

HomoloGene  
[24848](#), view the [multiple sequence alignment](#)

Ever confirmed somatic?  
Yes

FATHMM prediction  
Pathogenic (score 0.99)

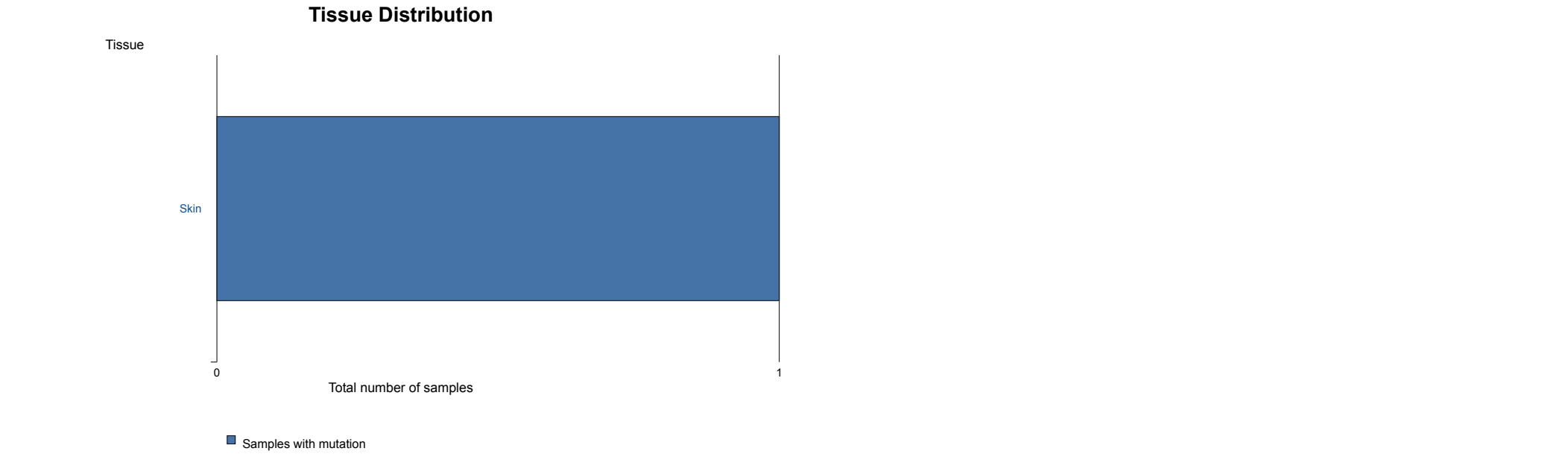
Remark  
n/a

Recurrent  
n/a

Drug resistance  
n/a

Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



Samples

This section displays a table of mutated samples, with tissue, histology and zygoty information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygoty	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">CSCC-37-T</a>	<a href="#">SEC14L3</a>	<a href="#">ENST00000215812</a>	<a href="#">Skin</a>	Head neck	Carcinoma	Squamous cell carcinoma	<a href="#">25303977</a>	Heterozygous	Confirmed Somatic	Tumour Sample	Unknown	-	

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References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
Mutational landscape of aggressive cutaneous squamous cell carcinoma	Pickering CR et al	2014	Clinical cancer research : an official journal of the American Association for Cancer Research	Curated	<a href="#">COSP37447</a>	<a href="#">25303977</a>

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Mutation



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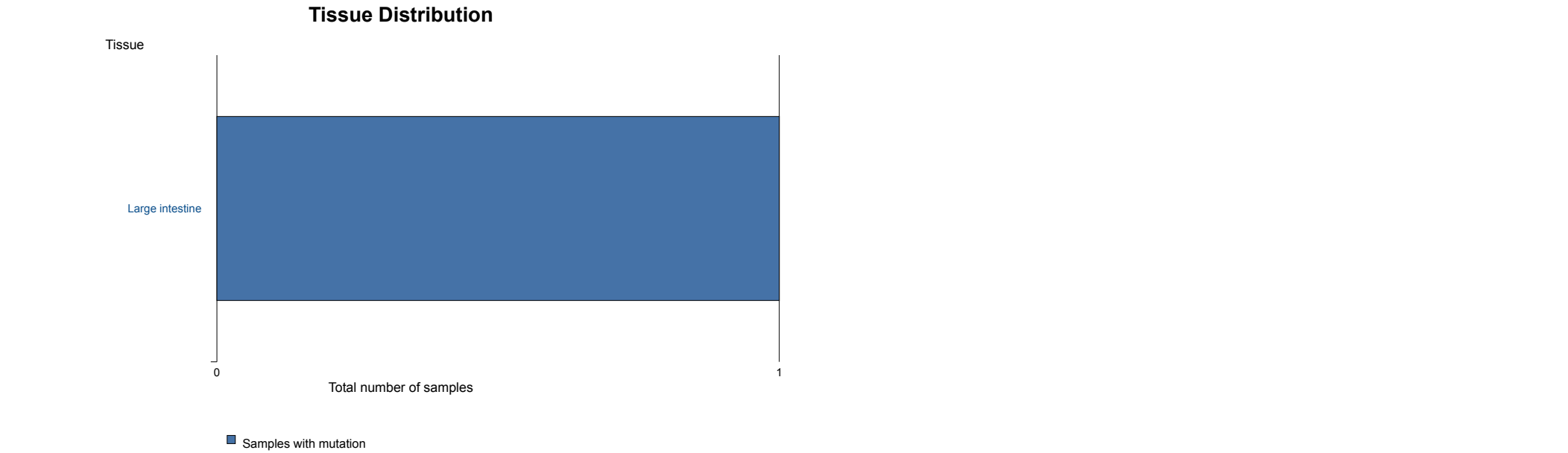
## Overview

This section shows a general overview of the selected mutation. It describes the source of the mutation i.e gene name/sample name/tissue name with unique ID, and also shows the mutation syntax at the amino acid and nucleotide sequence level. You can see more information on our [help pages](#).

Mutation ID	COSM6223153
Gene name	<a href="#">DDX3Y ENST00000537441</a>
AA mutation	p.R598Q (Substitution - Missense, position 598, R→Q)
CDS mutation	c.1793G>A (Substitution, position 1793, G→A)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">Y:12917441..12917441</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

## Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



Samples

This section displays a table of mutated samples, with tissue, histology and zygosity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygosity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">T073340A</a>	<a href="#">DDX3Y_ENST00000537441</a>	<a href="#">ENST00000537441</a>	<a href="#">Large intestine</a>	NS	Carcinoma	Adenocarcinoma	-	Unknown	Confirmed Somatic	Tumour Sample	Unknown	-	

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References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
ICGC(COCA-CN) : Colorectal Cancer - CN				Curated (Study)	<a href="#">COSU646</a>	

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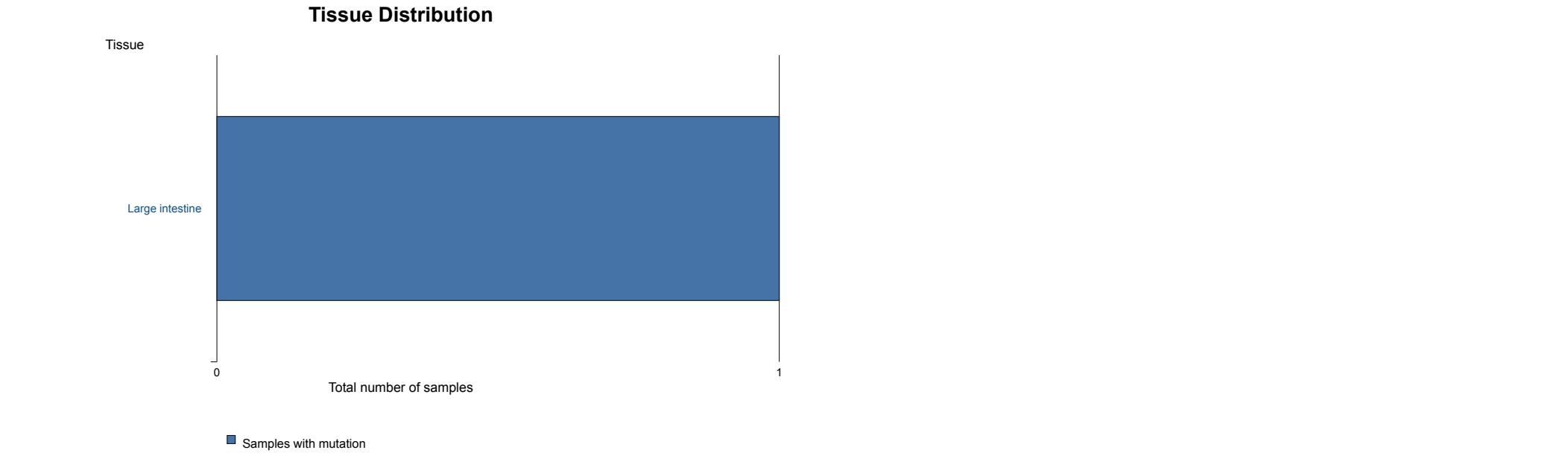
## Overview

This section shows a general overview of the selected mutation. It describes the source of the mutation i.e gene name/sample name/tissue name with unique ID, and also shows the mutation syntax at the amino acid and nucleotide sequence level. You can see more information on our [help pages](#).

Mutation ID	COSM6756630
Gene name	<a href="#">PRPF4B_ENST00000337659</a>
AA mutation	p.R650Q (Substitution - Missense, position 650, R→Q)
CDS mutation	c.1949G>A (Substitution, position 1949, G→A)
Nucleotides inserted	n/a
Genomic coordinates	GRCh38, <a href="#">6:4049029..4049029</a> , view <a href="#">Ensembl contig</a>
CDD	n/a
HomoloGene	n/a
Ever confirmed somatic?	Yes
FATHMM prediction	Pathogenic (score 0.99)
Remark	n/a
Recurrent	n/a
Drug resistance	n/a

## Tissue distribution

This section displays the distribution of mutated samples and tissue types (top 5). You can see more information on our [help pages](#).



### Samples

This section displays a table of mutated samples, with tissue, histology and zygosity information. Publication information is also included, where available, with links to PUBMED.

Sample name	Gene name	Transcript	Primary Tissue	Tissue Subtype 1	Primary Histology	Histology Subtype 1	Pubmed ID	Zygosity	Somatic Status	Sample Type	LOH	Resistant Mutation	Drugs
<a href="#">T253</a>	<a href="#">PRPF4B_ENST00000337659</a>	<a href="#">ENST00000337659</a>	<a href="#">Large intestine</a>	NS	Carcinoma	Adenocarcinoma	<a href="#">27149842</a>	Unknown	Confirmed Somatic		Unknown	-	

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### References

This section displays a table of references for the mutation. You can see more information on the [help pages](#).

Reference Title	Author	Year	Journal	Status	COSMIC	Pubmed
Genomic Correlates of Immune-Cell Infiltrates in Colorectal Carcinoma	Giannakis M et al	2016	Cell reports	Curated	<a href="#">COSP44021</a>	<a href="#">27149842</a>

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