

Assignment AL4: Precision Medicine Exploration

PART A

a.

Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withSNOMED CT US EditionNon-small cell lung cancer

① ↺ ↻ ↵

Description ↕	Alias Hit	SNOMED ID
Epidermal growth factor receptor negative non-small cell lung cancer	●	427038005
Epidermal growth factor receptor positive non-small cell lung cancer	●	426964009
Non-small cell adenocarcinoma of lung	●	1255725002
Non-small cell lung cancer		254637007
Non-small cell lung cancer with mutation in epidermal growth factor receptor		703228009
Non-small cell lung cancer without mutation in epidermal growth factor receptor		703230006
Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

① ↺

Select Content ModelSelect Code SetSelect Relationship

Name	Source Code	Has Qualifiers
Non-small cell lung cancer		has parent
Carcinoma of lung	448993007	
Non-small cell lung cancer		Has Child
Anaplastic lymphoma kinase fusion oncogene negative non-small cell lung cancer	830055006	
Anaplastic lymphoma kinase fusion oncogene positive non-small cell lung cancer	830151004	
Epidermal growth factor receptor negative non-small cell lung cancer	427038005	

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withSNOMED CT US EditionNon-small cell lung cancer

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100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

① ↺

Select Content ModelSelect Code SetSelect Relationship

Name	Source Code	Has Qualifiers
Epidermal growth factor receptor negative non-small cell lung cancer	427038005	
Epidermal growth factor receptor positive non-small cell lung cancer	426964009	
Non-small cell adenocarcinoma of lung	1255725002	
Non-small cell carcinoma of lung, TNM stage 1	424132000	
Non-small cell carcinoma of lung, TNM stage 2	425048006	
Non-small cell carcinoma of lung, TNM stage 3	422968005	

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Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

① ↺

Select Content ModelSelect Code SetSelect Relationship

Name	Source Code	Has Qualifiers
> Non-small cell carcinoma of lung, TNM stage 4	423121009	
> Non-small cell lung cancer with mutation in epidermal growth factor receptor	703228009	
> Non-small cell lung cancer without mutation in epidermal growth factor receptor	703230006	
> Non-small cell lung carcinoma with NRG1 fusion	1141627001	
> Nonsquamous nonsmall cell neoplasm of lung	440173001	
> Primary non-small cell lung cancer	1259727001	

- Reactive nitrogen species 1 negative non-small

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

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100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

① ↺

Select Content ModelSelect Code SetSelect Relationship

Name	Source Code	Has Qualifiers
> Reactive oxygen species 1 negative non-small cell lung cancer	830060005	
> Reactive oxygen species 1 positive non-small cell lung cancer	722425009	
> Squamous non-small cell lung cancer	723301009	
Non-small cell lung cancer ⊕ is a member of		
> General Practice / Family Practice reference set	450970008	
> Global Patient Set	787778008	

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Non-small cell lung carcinoma with NRG1 fusion1141627001

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Select Content ModelSelect Code SetSelect Relationship

NameSource CodeHas Qualifiers

> International Patient Summary816080008

Non-small cell lung cancer

SCT Associated morphology

> Non-small cell carcinoma128632008

Non-small cell lung cancer

SCT Finding site

> Lung structure39607008

Non-small cell lung cancer

SCT is associated finding

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Non-small cell lung carcinoma with NRG1 fusion1141627001

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Select Content ModelSelect Code SetSelect Relationship

NameSource CodeHas Qualifiers

> Lung structure39607008

Non-small cell lung cancer

SCT is associated finding

> History of bilateral lung non-small cell carcinoma16848221000119100

> History of non-small cell malignant neoplasm of lung429226001

> History of primary non-small cell carcinoma of left lung16848181000119108

> History of primary non-small cell carcinoma of right lung16848141000119103

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100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Aliases (3)

Alias	Code	Type	Language
Non-small cell lung cancer (disorder)	645558018	Fully Specified Name	English-US

Attributes for Non-small cell lung cancer (disorder)

Name	Value	Code
Is Preferred	1	
Module	SNOMED CT core	

Showing 2 alias attributes

> NSCLC - Non-small cell lung cancer379196015AlternateEnglish-US

> Non-small cell lung cancer379195016AlternateEnglish-US

Showing 3 aliases

Attributes (7)

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withSNOMED CT US EditionNon-small cell lung cancer

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Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Attributes (7)

Name	Value	Code
Active	1	
Code System OID	2.16.840.1.113883.6.96	
Code System URL	http://snomed.info/sct	
DefinitionStatus	900000000000073002	
Module	SNOMED CT core	
ModuleID	900000000000207008	
SemanticTag	disorder	

Mappings (2)

Code Set	Concept	Code	Retired	Target
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		✓
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		✓

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Term Search

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Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

ModuleSNOMED CT core

ModuleID900000000000207008

SemanticTagdisorder

Mappings (2)

Code Set ↕	Concept ↕	Code ↕	Retired ↕	Target ↕
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		●
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		●

Showing 2 mapping targets

External Codes (2)

Code ↕	Code Type ↕
4115276	OMOP
C0007131	UMLS CUI

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withSNOMED CT US EditionNon-small cell lung cancer

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Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

NSCLC - Non-small cell lung cancer379196015AlternateEnglish-US

Attributes for NSCLC - Non-small cell lung cancer

Name ↕	Value ↕	Code ↕
Is Preferred	0	
Module	SNOMED CT core	

Showing 2 alias attributes

Non-small cell lung cancer379195016AlternateEnglish-US

Showing 3 aliases

Attributes (7)

Additional information associated with the term		
Name ↕	Value ↕	Code ↕
Active	1	
Code System OID	2.16.840.1.113883.6.96	
Code System URL	http://snomed.info/sct	

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Term Search

Term begins with SNOMED CT US Edition Non-small cell lung cancer

Description <input type="text"/>	Alias Hit	SNOMED ID
Epidermal growth factor receptor negative non-small cell lung cancer	<input checked="" type="radio"/>	427038005
Epidermal growth factor receptor positive non-small cell lung cancer	<input checked="" type="radio"/>	426964009
Non-small cell adenocarcinoma of lung	<input checked="" type="radio"/>	1255725002
Non-small cell lung cancer	<input checked="" type="radio"/>	254637007
Non-small cell lung cancer with mutation in epidermal growth factor receptor	<input type="radio"/>	703228009
Non-small cell lung cancer without mutation in epidermal growth factor receptor	<input type="radio"/>	703230006
Non-small cell lung carcinoma with NRG1 fusion	<input checked="" type="radio"/>	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

Details Relationship Browser

Name <input type="text"/>	Value <input type="text"/>	Code <input type="text"/>
Active	1	
Code System OID	2.16.840.1.113883.6.96	
Code System URL	http://snomed.info/sct	
DefinitionStatus	900000000000073002	
Module	SNOMED CT core	
ModuleID	900000000000207008	
SemanticTag	disorder	

Mappings (2)

Code Set <input type="text"/>	Concept <input type="text"/>	Code <input type="text"/>	Retired <input type="text"/>	Target <input type="text"/>
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		<input checked="" type="checkbox"/>
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		<input checked="" type="checkbox"/>

Showing 2 mapping targets

Term Search

Term begins with SNOMED CT US Edition Non-small cell lung cancer

Description <input type="text"/>	Alias Hit	SNOMED ID
Epidermal growth factor receptor negative non-small cell lung cancer	<input checked="" type="radio"/>	427038005
Epidermal growth factor receptor positive non-small cell lung cancer	<input checked="" type="radio"/>	426964009
Non-small cell adenocarcinoma of lung	<input checked="" type="radio"/>	1255725002
Non-small cell lung cancer	<input checked="" type="radio"/>	254637007
Non-small cell lung cancer with mutation in epidermal growth factor receptor	<input type="radio"/>	703228009
Non-small cell lung cancer without mutation in epidermal growth factor receptor	<input type="radio"/>	703230006
Non-small cell lung carcinoma with NRG1 fusion	<input checked="" type="radio"/>	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

Details Relationship Browser

Module	SNOMED CT core
ModuleID	900000000000207008
SemanticTag	disorder

Mappings (2)

Code Set <input type="text"/>	Concept <input type="text"/>	Code <input type="text"/>	Retired <input type="text"/>	Target <input type="text"/>
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		<input checked="" type="checkbox"/>
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		<input checked="" type="checkbox"/>

Showing 2 mapping targets

External Codes (2)

Code <input type="text"/>	Code Type <input type="text"/>
4115276	OMOP
C0007131	UMLSCUI

Terminology Search

Taxonomy Browse

Terminology Mappings

Term Search

Term begins with

SNOMED CT US Edition

Non-small cell lung cancer

Q

X

▼

① ↺ ↻ ↴ ↵

Description ↕	Alias Hit	SNOMED ID
Epidermal growth factor receptor negative non-small cell lung cancer	●	427038005
Epidermal growth factor receptor positive non-small cell lung cancer	●	426964009
Non-small cell adenocarcinoma of lung	●	1255725002
Non-small cell lung cancer		254637007
Non-small cell lung cancer with mutation in epidermal growth factor receptor		703228009
Non-small cell lung cancer without mutation in epidermal growth factor receptor		703230006
Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100

<< < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

Details

Relationship Browser

Non-small cell lung cancer

379195016

Alternate

English-US

Attributes for Non-small cell lung cancer

Name ▼	Value ▼	Code ▼
Is Preferred	1	
Module	SNOMED CT core	

Showing 2 alias attributes

Showing 3 aliases

Attributes (7)

Additional information associated with the term		
Name ▼	Value ▼	Code ▼
Active	1	
Code System OID	2.16.840.1.113883.6.96	
Code System URL	http://snomed.info/sct	
DefinitionStatus	900000000000073002	

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Terminology Search

Taxonomy Browse

Terminology Mappings

Term Search

Term begins with

SNOMED CT US Edition

Non-small cell lung cancer

Q

X

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① ↺ ↻ ↴ ↵

Description ↕	Alias Hit	SNOMED ID
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Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100

<< < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

Details

Relationship Browser

Name ▼

Value ▼

Code ▼

Active

1

Code System OID

2.16.840.1.113883.6.96

Code System URL

http://snomed.info/sct

DefinitionStatus

900000000000073002

Module

SNOMED CT core

ModuleID

9000000000000207008

SemanticTag

disorder

Mappings (2)

Code Set ▼	Concept ▼	Code ▼	Retired ▼	Target ▼
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		●
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		●

Showing 2 mapping targets

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withSNOMED CT US EditionNon-small cell lung cancer

① ↺ ↻ ↴ ↵

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Non-small cell lung carcinoma with NRG1 fusion	●	1141627001

100 << < 1 > >>

Showing 1 to 7 of 7 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

ModuleSNOMED CT core

ModuleID900000000000207008

SemanticTagdisorder

Mappings (2)

Code Set	Concept	Code	Retired	Target
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		●
ICD-10-CM	Malignant neoplasm of unspecified part of unspecified bronchus or lung	C34.90		●

Showing 2 mapping targets

External Codes (2)

Code	Code Type
4115276	OMOP
C0007131	UMLS CUI

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b.

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withNCBI - MedGenNon-small cell lung cancer

① ↺ ↻ ↴ ↵

Term ↑	Alias Hit	Code
Non-small cell lung cancer		C0007131
Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201		C2827548
Non-small cell lung cancer with mutation in epidermal growth factor receptor		C3840002
Non-small cell lung cancer without mutation in epidermal growth factor receptor		C3838845

100 << < 1 > >>

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

> Aliases (0)

> Attributes (1)

Name	Value	Code
source	GTR	

Mappings (0)

Code Set	Concept	Code	Retired	Target
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Showing 0 mapping targets

External Codes (1)

Code	Code Type
254637007	SCT-ConceptID

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Term Search

Term begins with

Term	Alias Hit	Code
Non-small cell lung cancer		C0007131
Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201		C2827548
Non-small cell lung cancer with mutation in epidermal growth factor receptor		C3840002
Non-small cell lung cancer without mutation in epidermal growth factor receptor		C3838845

100 << < 1 > >>

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

Details Relationship Browser

 Select Content Model Select Code Set Select Relationship

Name	Source Code	Has Qualifiers
> NM_001374258.1(BRAF):c.1862A>G (p.Asn621Ser)	174177	
> NM_001374258.1(BRAF):c.1900G>A (p.Asp634Asn)	53980	
> NM_001374258.1(BRAF):c.1901A>G (p.Asp634Gly)	29011	
> NM_001374258.1(BRAF):c.1914_1916dup (p.Thr639dup)	176758	
> NM_001374258.1(BRAF):c.1918G>A (p.Val640Met)	53982	
> NM_001374258.1(BRAF):c.1918G>T (p.Val640Leu)	53983	

Term Search

Term begins with

Term	Alias Hit	Code
Non-small cell lung cancer		C0007131
Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201		C2827548
Non-small cell lung cancer with mutation in epidermal growth factor receptor		C3840002
Non-small cell lung cancer without mutation in epidermal growth factor receptor		C3838845

100 << < 1 > >>

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

Details Relationship Browser

 Select Content Model Select Code Set Select Relationship

Name	Source Code	Has Qualifiers
> NM_001374258.1(BRAF):c.1919_1921del (p.Val640_Lys641delinsGlu)	53984	
> NM_001374258.1(BRAF):c.1921A>G (p.Lys641Glu)	29005	
> NM_002524.5(NRAS):c.182A>G (p.Gln61Arg)	28939	
> NM_004448.4(ERBB2):c.2313_2324dup (p.Tyr772_Ala775dup)	28914	
> NM_004448.4(ERBB2):c.2314_2325dup (p.Tyr772_Ala775dup)	54152	
> NM_004448.4(ERBB2):c.2320del (p.Met774fs)	54153	
> NM_004448.4(ERBB2):c.2324_2325insCTCCGTGA1	176403	

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withNCBI - MedGenNon-small cell lung cancer

Term

Alias Hit

Code

Non-small cell lung cancer

C0007131

Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201

C2827548

Non-small cell lung cancer with mutation in epidermal growth factor receptor

C3840002

Non-small cell lung cancer without mutation in epidermal growth factor receptor

C3838845

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Select Content Model

Select Code Set

Select Relationship

Name

Source Code

Has Qualifiers

NM_004448.4(ERBB2):c.2324_2325insTCCGIGAI (p.Gly776_Val777InsSerValMetAla)

176493

NM_004448.4(ERBB2):c.2325_2326insTCCGTGATC (p.Gly776_Val777InsSerValMetAla)

176631

NM_004448.4(ERBB2):c.2326_2327insTGT (p.Gly776delInsValCys)

54155

NM_004448.4(ERBB2):c.2326_2327insTTT (p.Gly776delInsValCys)

54156

NM_004448.4(ERBB2):c.2326delInsTTAT (p.Gly776delInsLeuCys)

54157

NM_004448.4(ERBB2):c.2331_2339dup (p.Gly778_Pro780dup)

54159

NM_004448.4(ERBB2):c.2332_2340dup

54160

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withNCBI - MedGenNon-small cell lung cancer

Term

Alias Hit

Code

Non-small cell lung cancer

C0007131

Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201

C2827548

Non-small cell lung cancer with mutation in epidermal growth factor receptor

C3840002

Non-small cell lung cancer without mutation in epidermal growth factor receptor

C3838845

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Select Content Model

Select Code Set

Select Relationship

Name

Source Code

Has Qualifiers

NM_004448.4(ERBB2):c.2332_2340dup (p.Gly778_Pro780dup)

54160

NM_004985.5(KRAS):c.181C>A (p.Gln61Lys)

175715

NM_004985.5(KRAS):c.182A>T (p.Gln61Leu)

54283

NM_004985.5(KRAS):c.34G>A (p.Gly12Ser)

27623

NM_004985.5(KRAS):c.37G>C (p.Gly13Arg)

27632

NM_004985.5(KRAS):c.37G>T (p.Gly13Cys)

54290

NM_004985.5(KRAS):c.38G>A (p.Gly13Asp)

27619

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Term Search

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Term

Alias Hit

Code

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Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201

C2827548

Non-small cell lung cancer with mutation in epidermal growth factor receptor

C3840002

Non-small cell lung cancer without mutation in epidermal growth factor receptor

C3838845

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Select Content Model

Select Code Set

Select Relationship

Name

Source Code

Has Qualifiers

> NM_005228.5(EGFR):c.2303_2304delinsTT (p.Ser768Ile)

54419

> NM_005228.5(EGFR):c.2303G>T (p.Ser768Ile)

54418

> NM_005228.5(EGFR):c.2312_2314dup (p.Pro772_His773insHis)

54425

> NM_005228.5(EGFR):c.2315_2320dup (p.Val774_Cys775insAlaHis)

54427

> NM_005228.5(EGFR):c.2500G>T (p.Val834Leu)

54446

> NM_006218.4(PIK3CA):c.1359_1373del (p.Glu453_Pro458delinsAsp)

54632

> NM_006218.4(PIK3CA):c.3073A>G

54634

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edu.symedical.com/viewpoint/vpc/termsearch

Symedical® Viewpoint

Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withNCBI - MedGenNon-small cell lung cancer

Term

Alias Hit

Code

Non-small cell lung cancer

C0007131

Non-Small Cell Lung Cancer mRNA-Derived Vaccine CV9201

C2827548

Non-small cell lung cancer with mutation in epidermal growth factor receptor

C3840002

Non-small cell lung cancer without mutation in epidermal growth factor receptor

C3838845

Showing 1 to 4 of 4 terms

Term Detail: Non-small cell lung cancer

DetailsRelationship Browser

Select Content Model

Select Code Set

Select Relationship

Name

Source Code

Has Qualifiers

> NM_005228.5(EGFR):c.2315_2320dup (p.Val774_Cys775insAlaHis)

54427

> NM_005228.5(EGFR):c.2500G>T (p.Val834Leu)

54446

> NM_006218.4(PIK3CA):c.1359_1373del (p.Glu453_Pro458delinsAsp)

54632

> NM_006218.4(PIK3CA):c.3073A>G (p.Thr1025Ala)

54634

> NM_006218.4(PIK3CA):c.3129G>A (p.Met1043Ile)

173901

> NM_006218.4(PIK3CA):c.3139C>T (p.His1047Tyr)

48304

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PART B

Symedical® Viewpoint

Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Word begins withSNOMED CT US Editionmutation

Description

Alias Hit

SNOMED ID

Activated PI3K-delta syndrome		711480000
Acute infantile liver failure with multisystemic involvement syndrome		774207004
Acute myeloid leukemia with biallelic CEBPA mutation		788740009
Acute myeloid leukemia with CEBPA mutation		703819004
Acute myeloid leukemia with CEBPA somatic mutations		764855007
Acute myeloid leukemia with NPM1 somatic mutation		763309005
Acute neuropathic Gaucher's disease		12246008
Acyl-CoA dehydrogenase 9 deficiency		725046003
ADan amyloidosis		783258000
ADNP-related multiple congenital anomalies, intellectual disability, autism spectrum disorder		766824003
Adult polyglucosan body disease		721099001

Showing 1 to 100 of 1139 terms

Term Detail: Acute myeloid leukemia with CEBPA mutation

DetailsRelationship Browser

Select Content Model

Select Code Set

Select Relationship

Name	Source Code	Has Qualifiers
Acute myeloid leukemia with CEBPA mutation		has parent
Acute myeloid leukemia	1162928000	
Acute myeloid leukemia with CEBPA mutation		Has Child
Acute myeloid leukemia with biallelic CEBPA mutation	788740009	
Acute myeloid leukemia with CEBPA mutation		SCT is associated morphology
Acute myeloid leukemia with CEBPA somatic mutations	764855007	

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Terminology SearchTaxonomy BrowseTerminology Mappings

Term Search

Term begins withClinVar - Variant - MPM ...nm cebpa

Term

Alias Hit

Code

NM_000104.4(CYP1B1):c.*975A>G		286953
NM_000104.4(CYP1B1):c.*980A>C		884837
NM_000104.4(CYP1B1):c.*981T>C		286952
NM_000104.4(CYP1B1):c.1043+275C>T		1196920
NM_000104.4(CYP1B1):c.1043+6G>C		960477
NM_000104.4(CYP1B1):c.1044-147dup		1210929
NM_000104.4(CYP1B1):c.1044-3C>G		962068
NM_000104.4(CYP1B1):c.1047T>C (p.Tyr349=)		691155
NM_000104.4(CYP1B1):c.1062_1075del (p.Arg355fs)		404760
NM_000104.4(CYP1B1):c.1064_1076del (p.Arg355fs)		266801
NM_000104.4(CYP1B1):c.1078T>C (p.Leu360=)		691154
NM_000104.4(CYP1B1):c.1081G>T (p.Asp361Tyr)		1053909

Showing 1 to 100 of 2168 terms

Term Detail: NM_000104.4(CYP1B1):c.1064_1076del (p.Arg355fs)

DetailsRelationship Browser

Select Content Model

Select Code Set

Select Relationship

Name	Source Code	Has Qualifiers
NM_000104.4(CYP1B1):c.1064_1076del (p.Arg355fs)		PMM - Has Associated Phenotype
Developmental glaucoma	HP:0001087	
NM_000104.4(CYP1B1):c.1064_1076del (p.Arg355fs)		PMM - Has Associated Phenotype
Congenital glaucoma	C0020302	
not provided	CNS17202	

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PART C

C2a.

SNOMED CT, a standardized clinical terminology, offers a broad vocabulary for recording detailed medical data across various contexts, including disease diagnosis, treatment procedures, and patient outcomes (Chang & Mostafa, 2021). Its comprehensive structure allows for precise clinical documentation and interoperability across health systems (Chang & Mostafa, 2021).

On the other hand, NCBI – MedGen focuses on genetic disorders, providing in-depth information about phenotypes, inheritance patterns, and the genetic underpinnings of diseases

(NCBI, n.d.). It links to genetic databases and literature, facilitating research and clinical understanding of genetic conditions ((MedGen Help Documentation, n.d.)

b. The overlap between these two systems enhances patient care by integrating detailed clinical descriptions with genetic insights. This integration supports personalized medicine, allowing for targeted treatments based on a patient's specific clinical presentation and genetic profile(Chang & Mostafa, 2021). However, gaps in this integration could limit the effectiveness of such personalized approaches, underscoring the importance of ongoing efforts to link clinical and genetic data more closely.

C4a.

SNOMED CT functions as a standardized framework for documenting and reporting clinical healthcare information. It encompasses a vast range of medical terms, encompassing diagnoses, procedures, medications, allergies, and outcomes. This comprehensive structure facilitates communication and data sharing across diverse healthcare settings, ensuring detailed and accurate representation of patient data (SNOMED International, n.d.).

On the other hand, ClinVar focuses specifically on the genetic aspect of healthcare, cataloging information about genomic variation and its impact on human health (Landrum et al., 2015). It includes detailed annotations and interpretations of genetic variants, offering valuable insights into their relevance to various diseases. This information is crucial for geneticists, researchers, and clinicians in diagnosing genetic conditions, understanding variant pathogenicity, and guiding treatment decisions informed by genetic data (Landrum et al., 2015).

b.The relationship data in ClinVar plays a significant role in personalized medicine for individuals with genetic variants. It provides essential information for diagnosis, prognostic assessments, and identifying potential therapeutic options (Landrum et al., 2015). However, the absence of clear, annotated relationships or interpretations for some variants creates uncertainties in clinical decision-making processes. This underscores the importance of comprehensive genetic data analysis for advancing personalized medicine.

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