

[PORTAL-2] Decide on disease list that pipeline supports, assign corresponding tissues and implement in pipeline and portal

Created: 03/Feb/21 Updated: 26/Dec/22 Due: 05/Oct/22

Status:	Reopened
Project:	Portal
Components:	None
Affects versions:	None
Fix versions:	None

Type:	Task	Priority:	Highest
Reporter:	Philipp Anokhin	Assignee:	Dhaval Patel
Resolution:	Unresolved	Votes:	0
Labels:	None		
Remaining Estimate:	Not Specified		
Time Spent:	Not Specified		
Original estimate:	Not Specified		

Attachments:	Disease.xlsx	image-20210626-064123.png	image-20210626-064142.png
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Description

UPDATE: Dhaval Patel to implement the list in portal.

UPDATE2: Here is a mapping file for EFO (used by OpenTargets – in the Disease column below) → ICD10 (used for accession number generation) mappings (not all terms are mapped though!): https://github.com/EBISpot/EFO-UKB-mappings/blob/master/UK_Biobank_master_file.tsv

UPDATE3: Search for disease names at <https://www.icd10data.com/ICD10CM/Index/A/Adenocarcinoma> and map disease names to general ICD10 codes. E.g. Adenocarcinoma maps to

UPDATE4: Here is a EFO->ICD10(Malacard) mapping file from OpenTargets project itself:
<https://docs.google.com/spreadsheets/d/1GSOkYegyQOd21gdhMEex6gkOsHzlF9DWApg096FIo8>

When doctor creates an account for a patient, they should select tissue name (location of the disease) in addition to a disease name. Please display a drop-down with a disease first and, after the doctor chooses a disease, display relevant tissues as radio buttons.

Store both disease and tissue names in DB for retrieval by pipeline.

Here are the **final** disease/tissue mappings for the drop-downs (use as parameters to call Jenkins pipeline and in portal):

Tissue	Disease	EFO disease code
AdiposeSubcutaneous	Liposarcoma	EFO_0000569
AdiposeVisceral	MalignantPleuralMesothelioma	EFO_0000770
BrainCortex	AdrenalCortexNeoplasm	EFO: MONDO_0036591
Skin <-Not sun exposed!	SkinNeoplasm	EFO: EFO_0004198
SkinSun	Melanoma	EFO_0000756
	BasalCellCarcinoma	EFO_0004193
	SkinSquamousCellCarcinoma	MONDO_0002529
	MerkelCellSkinCancer	EFO_1001471
	KaposiSarcoma	EFO_0000558
	Angiosarcoma	EFO_0003968
Ovary	OvarianCancer	EFO: MONDO_0008170
NerveTibial	NeurofibromatosisType1	EFO: Orphanet_636
	tibial neuropathy	EFO: EFO_1001243 ← This one is not good, only 1 result from OpenTargets
Esophagus	NeoplasmOfEsophagus	EFO: MONDO_0021355
	BarrettEsophagus	EFO: EFO_0000280
	esophageal cancer	EFO: MONDO_0007576 ← results included in "neoplasm of esophagus"
	EsophagealAdenocarcinoma	EFO_0000478
	EsophagealCarcinoma	EFO: EFO_0002916
AdrenalGland	AdrenalGlandNeoplasm	EFO: EFO_0003850
	adrenal gland cancer	EFO: MONDO_0002847
	AdrenalGlandNeuroblastoma	EFO: EFO_1000075
MinorSalivaryGland	MinorSalivaryGlandAdenocarcinoma	EFO: EFO_1000379
	Malignant tumor of minor salivary gland	EFO: MONDO_0021346
	NeoplasmOfMinorSalivaryGland	EFO: MONDO_0021370
	SalivaryGlandCarcinoma	EFO: MONDO_0000521
Spleen	SpleenNeoplasm	EFO: MONDO_0036696
	spleen cancer	EFO: EFO_0007494

Thyroid	ThyroidNeoplasm thyroid cancer	EFO: EFO_0003841 EFO: MONDO_0002408
Vagina	VaginalNeoplasm VaginalMelanoma	EFO: EFO_1001447 EFO: EFO_1000619
FallopianTube	FallopianTubeNeoplasm	EFO: MONDO_0021092
Prostate	ProstateNeoplasm ProstateAdenocarcinoma Adenocarcinoma UrothelialCarcinoma ProstateNeuroendocrineNeoplasm SmallCellCarcinoma MetastaticProstateCancer	MONDO_0021259 EFO_0000673 EFO_0000228 EFO_0008528 MONDO_0002477 EFO_0008524 EFO_0000196
Pituitary	PituitaryTumor	MONDO_0017611
BrainCerebellum	Glioma GlioblastomaMultiforme Meningioma Craniopharyngioma Astrocytoma Ependymoma Oligodendroglioma GerminomatousGermCellTumor Medulloblastoma LeptomeningealMetastasis	EFO_0005543 EFO_0000519 Orphanet_2495 EFO_1000209 EFO_0000272 EFO_1000028 EFO_0000632 MONDO_0020580 EFO_0002939 EFO_1001012
Breast	BreastNeoplasm TripleNegativeBreastCancer InvasiveBreastDuctalCarcinoma InvasiveLobularCarcinoma InflammatoryBreastCarcinoma Angiosarcoma PhyllodesTumor LeptomeningealMetastasis BreastDuctalCarcinomaInsitu	EFO_0003869 EFO_0005537 EFO_0000186 EFO_0000553 EFO_1000984 EFO_0003968 EFO_0000653 EFO_1001012 EFO_0000432
ColonSigmoid ColonTransverse SmallIntestine	IntestinalNeoplasm ColorectalNeoplasm ColorectalAdenocarcinoma CarcinoidTumor GastrointestinalStromalTumor Leiomyosarcoma MalignantColonNeoplasm	MONDO_0021118 EFO_0004142 EFO_0000365 EFO_0004243 Orphanet_44890 EFO_0000564 MONDO_0021063
LymphNodes	FollicularLymphoma	MONDO_0018906
MuscleSkeletal	Rhabdomyosarcoma EpithelioidSarcoma Osteosarcoma	EFO_0002918 MONDO_0017387 EFO_0000637
Liver	LiverNeoplasm HepatocellularCarcinoma FibrolamellarCarcinoma Cholangiocarcinoma Angiosarcoma	EFO_1001513 EFO_0000182 EFO_1000256 EFO_0005221 EFO_0003968
Lung	LungNeoplasm LungAdenocarcinoma NonSmallCellLungCarcinoma SmallCellLungCarcinoma LungCarcinoidTumor AdenosquamousLungCarcinoma SquamousCellLungCarcinoma LargeCellNeuroendocrineCarcinoma AdenoidCysticCarcinoma MucoepidermoidCarcinoma Mesothelioma	MONDO_0021117 EFO_0000571 EFO_0003060 EFO_0000702 EFO_1000037 EFO_0000233 EFO_0000708 EFO_0000563 EFO_0000231 MONDO_0003036 EFO_0000588
KidneyCortex KidneyMedulla	KidneyNeoplasm RenalCellCarcinoma TransitionalCellCarcinoma KidneySarcoma	EFO_0003865 EFO_0000681 EFO_1000601 MONDO_0002930
Bladder	BladderTumor PapillaryCarcinoma	EFO_0000294 EFO_1000646

	BladderAdenocarcinoma	EFO_1000125
Stomach	StomachNeoplasm	EFO_0003897
	GastricAdenocarcinoma	EFO_0000503
	GastricNonCardiaCarcinoma	EFO_0008502
	CarcinoidTumor	EFO_0004243
Uterus	UterineNeoplasm	EFO_0003859
	UterineCancer	MONDO_0002715
	EndometrialCancer	MONDO_0011962
	UterineSarcoma	EFO_0002914
	SerousAdenocarcinoma	EFO_0003825
	AdenosquamousCarcinoma	EFO_1000073
	UterineCarcinosarcoma	EFO_1000613
Testis	TesticularNeoplasm	EFO_0004281
	Seminoma	MONDO_0003001
	TesticularNonSeminomatousGermCellTumor	EFO_1000570
	MalignantTesticularGermCellTumor	MONDO_0003510
Pancreas	PancreaticNeoplasm	EFO_0003860
	PancreaticAdenocarcinoma	EFO_1000044
	PancreaticNeuroendocrineTumor	EFO_1000045
	MalignantExocrinePancreasNeoplasm	MONDO_0002116
EsophagusMucosa	EsophagealSquamousCellCarcinoma	EFO_0005922
	EsophagealAdenocarcinoma	EFO_0000478
EsophagusGastroesophagealJunction	EsophagealAdenocarcinoma	EFO_0000478

Work-in-progress disease list:

Tissue	Disease
Prostate	<p>ProstateNeoplasm</p> <p>Adenocarcinoma</p> <p>Acinar adenocarcinoma (conventional adenocarcinoma) ? (multiple possible matches in OT): Acinar Prostate Adenocarcinoma, Foamy Gland Variant</p> <p>EFO: EFO_1000064 <i>This category only has 4 hits in OT - doesn't make sense to use in pipeline</i></p> <p>Prostatic ductal adenocarcinoma (PDA) ? (should be in breast?)</p> <p>Transitional cell carcinoma OR UrothelialCarcinoma ? Both OK</p> <p>Neuroendocrine tumors, or carcinoids ? (no hits in OT for prostate): ProstateNeuroendocrineNeoplasm. EFO: MONDO_0002477</p> <p>Small cell carcinoma ? (should be in lung?) SmallCellCarcinoma EFO: EFO_0008524. this is neuroendocrine carcinoma prostate tissue. also in lung tissue.</p> <p>Squamous cell carcinoma ? (not for prostate)</p> <p>Prostate sarcoma ? (only 8 targets!) Ok Shamsuddin Sultan Khan, 8 targets will almost never be selected by pipeline - it doesn't make sense to use this category with so few targets</p> <p>Recurrent prostate cancer ? (no hits in OT)</p> <p>MetastaticProstateCancer</p>
Pituitary	PituitaryTumor
BrainCerebellum	<p>Glioma</p> <p>glioblastoma multiforme</p> <p>meningioma</p> <p>Craniopharyngioma (only 210 hits in OT)</p> <p>Astrocytoma</p> <p>Ependymoma</p> <p>Oligodendroglioma</p> <p>Germinoma ? (GerminomatousGermCellTumor OR germ cell tumor?) all ok</p> <p>Pineal region tumor ? (pineal body neoplasm OR pineal gland cancer? or Papillary Tumor of the Pineal Region) low hits from OT <i>This category only has 3 hits in OT - doesn't make sense to use in pipeline</i></p> <p>Medulloblastoma</p>
Breast	<p>BreastNeoplasm</p> <p>Triple-negative breast cancer</p> <p>Metastatic breast cancer ? (not found in OT)</p> <p>InvasiveBreastDuctalCarcinoma</p> <p>InvasiveLobularCarcinoma (458 hits in OT)</p> <p>InflammatoryBreastCarcinoma</p> <p>Page't's disease of the breast ? (no breast-specific Paget disease in OT)</p> <p>Angiosarcoma of the breast ? (can also be in skin, not specific in OT)</p> <p>PhyllodesTumor (261 hit in OT)</p>
ColonSigmoid	IntestinalNeoplasm
ColonTransverse	ColorectalAdenocarcinoma
SmallIntestine	<p>CarcinoidTumor (616 hits in OT)</p> <p>Primary colorectal lymphoma ? (only 12 hits in OT) Ok With only 12 hits in OT it doesn't make sense to use in pipeline</p>

	GastrointestinalStromalTumor Leiomyosarcoma (839 hits in OT)
SkinSun	Melanoma Recurrent BasalCellCarcinoma SkinSquamousCellCarcinoma Merkel-cell-carcinoma MerkelCellSkinCancer KaposiSarcoma Angiosarcoma Lymphoma of the skin, or cutaneous lymphoma ? (multiple variants available: primary, t-cell, follicular - which one do we pick?) Based on Diagnosis info I need a concrete answer here: do we use all, some, none of these 3 for skin tissue?
LymphNodes	FollicularLymphoma
MuscleSkeletal	Rhabdomyosarcoma
Liver	LiverNeoplasm HepatocellularCarcinoma Fibrolamellar (only 123 hits in OT) Cholangiocarcinoma Angiosarcoma
Lung	LungNeoplasm LungAdenocarcinoma NonSmallCellLungCarcinoma SmallCellLungCarcinoma LungCarcinoidTumor (only 229 hits in OT) AdenosquamousLungCarcinoma (only 357 hits in OT) SquamousCellLungCarcinoma LargeCellNeuroendocrineCarcinoma (only 278 hits in OT) Salivary-gland-type-lung-carcinoma ? AdenoidCysticCarcinoma EFO: EFO_0000231 MucoepidermoidCarcinoma EFO: MONDO_0003036 Mesothelioma
KidneyCortex KidneyMedulla	KidneyNeoplasm RenalCellCarcinoma TransitionalCellCarcinoma KidneySarcoma (only 109 hits in OT) Ok
Bladder	BladderTumor Papillary carcinoma Flat carcinomas ? (no hits in OT) BladderAdenocarcinoma (only 86 hits in OT) Ok bladder sarcoma ? (only 2 hits in OT) Can consider Bladder Tumor BladderTumor already included above - removing
Stomach	StomachNeoplasm GastricAdenocarcinoma Gastric carcinoid ? (not in OT) Its GastricNonCardiaCarcinoma EFO: EFO_0008502 CarcinoidTumor (616 hits in OT)
Uterus	UterineNeoplasm UterineCancer Endometrial cancer UterineSarcoma (346 hits in OT) SerousAdenocarcinoma AdenosquamousCarcinoma (646 hits in OT) UterineCarcinosarcoma (711 hits in OT)
Testis	TesticularNeoplasm Seminoma (833 hits in OT) TesticularNonSeminomatousGermCellTumor (146 hits in OT) spermatocytic seminoma ? (only 64 hits in OT) Ok Recurrent testicular cancer ? or can consider Testicular Neoplasm TesticularNeoplasm already included above - removing
Pancreas	PancreaticNeoplasm PancreaticAdenocarcinoma Endocrine tumor ? (not in OT for pancreas) or PancreaticNeuroendocrineTumor EFO: EFO_1000045 MalignantExocrinePancreasNeoplasm
EsophagusMucosa	EsophagealSquamousCellCarcinoma EsophagealAdenocarcinoma Squamous cell carcinoma ? (no esophagus-specific squamous cell carcinoma in OT) or esophageal squamous cell carcinoma EFO: EFO_0005022 already included above - removing

EsophagusGastroesophagealJunction	EsophagealAdenocarcinoma
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Please see link for all cancer types:

<https://www.cancer.gov/types#a>

Comments

Comment by [Dhaval Patel](#) [17/Mar/21]

Its implemented

Comment by [Dhaval Patel](#) [30/Apr/21]

[Philipp Anokhin](#) , is there any relationship between Tissue and Disease?

Comment by [Philipp Anokhin](#) [30/Apr/21]

Currently it's 1 to 1. First the doctor should select a disease and then tissue (tissue list should be auto-populated depending on the disease). Later on we may have several

Comment by [Philipp Anokhin](#) [11/May/21]

[Dhaval Patel](#) , please note the updated tissue/disease mapping table. Also, now there are several tissues per disease for Intestinal Neoplasm and Kidney Neoplasm.

Comment by [Philipp Anokhin](#) [15/May/21]

[john Catanzaro](#) , please take a look at the current list of disease/tissue mappings and let us know if we need to add more. Below are the tissues for which we have data but disease:

```
{'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Adipose - Subcutaneous.tsv'      'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Adipose - Visceral (Omentum).tsv'
'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Nerve - Tibial.tsv' }}
{'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Adipose - Visceral (Omentum).tsv' 'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Adipose - Visceral (Omentum).tsv'
GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Ovary.tsv }}
{'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Adrenal Gland.tsv'              'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Adrenal Gland.tsv'
GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Pituitary.tsv }}
{'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Artery - Aorta.tsv'            'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Artery - Aorta.tsv'
'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Skin - Not Sun Exposed (Suprapubic).tsv' }}
{'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Artery - Coronary.tsv'          'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Artery - Coronary.tsv'
GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Spleen.tsv }}
{'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Artery - Tibial.tsv'           'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Artery - Tibial.tsv'
GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Thyroid.tsv }}
'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Brain - Cortex.tsv'              'GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Brain - Cortex.tsv'
Salivary Gland.tsv'                          GTEx_Analysis_2017-06-05_v8_RNASeQCv1.1.9_gene_reads-Vagina.tsv
```

Comment by [john Catanzaro](#) [15/May/21]

Got it

Comment by [Stuart Ross](#) [26/Jun/21]

Tissue field needs to be populated. There is nothing in the drop downs

Comment by [Dhaval Patel](#) [26/Jun/21]

[Stuart Ross](#) , We have functionality into admin portal to add new disease and tissues as well,

here you can add new disease

PORTAL360

Dashboard

Master

Role

Disease

Portal Module

Role Module Mapping

Organization User

Neoantigen

Tissue

Patient

Patients

Payment

Home / Master / Disease

Disease List

Active

Search...

Disease Name	Disease Code	Status	Action
Testicular Neoplasm	TN	Active	Edit
Liver Neoplasm	LN	Active	Edit
Intestinal Neoplasm	IN	Active	Edit
Esophageal Adenocarcinoma	EA	Active	Edit
Esophageal Squamous Cell Carinoma	ESCC	Active	Edit
Bladder Tumor	BT	Active	Edit

and for tissue,

PORTAL360

Dashboard

Master

Role

Disease

Portal Module

Role Module Mapping

Organization User

Neoantigen

Tissue

Patient

Patients

Payment

Home / Master / Tissue

Tissue List

Active

Search...

Tissue Name	Disease	Status
Amyotrophic	Amyotrophic Lateral Sclerosis 1 (ALS1)	Active
Colorectal	Colorectal Cancer (CRC)	Active
Rectum	Rectum Cancer	Active
SmallIntestine	Intestinal Neoplasm	Active
KidneyMedulla	Kindney Neoplasm	Active
EsophagusGastroesophagealJunction	Esophageal Adenocarcinoma	Active

While creating tissues , you need to select specific disease.

Its master database for Disease and tissues .

Please let me know still you have any doubts.

I will give you proper demo about how to use this functionality.

Comment by [Stuart Ross](#) [28/Jun/21]

I still pick a disease but no Tissue type is available in the drop down

Comment by [John Catanzaro](#) [06/Jul/21]

[Philipp Anokhin](#) see <https://www.cancer.gov/types#a>

Comment by [John Catanzaro](#) [06/Jul/21]

For all cancer types needed to be uploaded.

Comment by [John Catanzaro](#) [06/Jul/21]

[Philipp Anokhin](#) all of the cancers listed in this data set are the main categories for all cancers

Comment by [John Catanzaro](#) [06/Jul/21]

[Philipp Anokhin Dhaval Patel](#)

<https://www.genome.jp/kegg/disease/>

Comment by [John Catanzaro](#) [06/Jul/21]

Disease classification Kegg

Comment by [John Catanzaro](#) [06/Jul/21]

[Dhaval Patel](#)

Comment by [John Catanzaro](#) [06/Jul/21]

[Philipp Anokhin Dhaval Patel](#) see <https://www.cancer.gov/types#a>

Comment by [Philipp Anokhin](#) [07/Jul/21]

[John Catanzaro](#), current Cancer pipeline uses OpenTargets for gene-to-disease associations and EFO (Experimental Factor Ontology): https://www.ebi.ac.uk/ols/ontologies/iri=http%3A%2F%2Fwww.ebi.ac.uk%2Fefo%2FEFO_0000408&viewMode=All&siblings=false for disease classification (since OpenTargets use EFO). You can see differences e.g. Breast Neoplasm and triple-negative breast cancer here: https://platform.opentargets.org/disease/EFO_0003869/associations and https://platform.opentargets.org/disease/EFO_0005537/associations.

Note that currently I use "Breast Neoplasm" for all kinds of breast cancer (including triple-negative). That was based on early recommendations from Anton Yuryev to take no evidence from literature.

Comment by [John Catanzaro](#) [13/Jul/21]

[Philipp Anokhin](#) starting Cancer Disease Ontology List: <https://docs.google.com/document/d/1OhLZMtG66RYLvEVCjPPmBSpnTeX8OEyY5oITPc4YoSs/edit>

Comment by [John Catanzaro](#) [13/Jul/21]

[Shamsuddin Sultan Khan](#) <https://docs.google.com/document/d/1OhLZMtG66RYLvEVCjPPmBSpnTeX8OEyY5oITPc4YoSs/edit>

Comment by [John Catanzaro](#) [13/Jul/21]

[Shamsuddin Sultan Khan](#) Building Ontology Disease List Link above

Comment by [John Catanzaro](#) [13/Jul/21]

[Philipp Anokhin Shamsuddin Sultan Khan Dhaval Patel](#) I will continue to build disease ontology list. Got a good jump going

Comment by [John Catanzaro](#) [13/Jul/21]

[Dhaval Patel](#) <https://docs.google.com/document/d/1OhLZMtG66RYLvEVCjPPmBSpnTeX8OEyY5oITPc4YoSs/edit>

Comment by [Shamsuddin Sultan Khan](#) [20/Jul/21]

I've started work on compiling disease names for PES

Comment by [Shamsuddin Sultan Khan](#) [23/Jul/21]

Please see below disease list:

Tissue	Disease
Prostate	ProstateNeoplasm Adenocarcinoma Acinar adenocarcinoma (conventional adenocarcinoma) ? Prostatic ductal adenocarcinoma (PDA) ? (should be in breast?) Transitional cell carcinoma OR urothelial carcinoma ? Neuroendocrine tumors, or carcinoids ? (no hits in OT for prostate) Small cell carcinoma ? (should be in lung?) Squamous cell carcinoma ? (not for prostate) Prostate sarcoma (only 8 targets!) Recurrent prostate cancer ? (no hits in OT) Metastatic prostate cancer
BrainCerebellum	Glioma glioblastoma multiforme meningioma craniopharyngioma (only 210 hits in OT) pituitary tumor Astrocytoma Ependymoma Oligodendroglioma Craniopharyngioma (only 210 hits in OT) Germinoma ? (germinomatous germ cell tumor OR germ cell tumor?) Pineal region tumor ? (pineal body neoplasm OR pineal gland cancer?) low hits from OT Medulloblastoma

Breast	BreastNeoplasm Triple-negative breast cancer Metastatic breast cancer ? (not found in OT) InvasiveBreastDuctalCarcinoma InvasiveLobularCarcinoma (458 hits in OT) InflammatoryBreastCarcinoma Paget's disease of the breast (no breast-specific Paget disease in OT) Angiosarcoma of the breast ? (can also be in skin, not specific in OT) PhyllodesTumor (261 hit in OT)
ColonSigmoid ColonTransverse SmallIntestine	IntestinalNeoplasm ColorectalAdenocarcinoma CarcinoidTumor (616 hits in OT) Primary colorectal lymphoma ? (only 12 hits in OT) GastrointestinalStromalTumor Leiomyosarcoma (839 hits in OT)
SkinSun	Melanoma Recurrent basal cell carcinoma Squamous cell carcinoma Merkel cell carcinoma Kaposi sarcoma Lymphoma of the skin, or cutaneous lymphoma
LymphNodes	FollicularLymphoma
MuscleSkeletal	Rhabdomyosarcoma
Liver	LiverNeoplasm Hepatocellular carcinoma Fibrolamellar Cholangiocarcinoma Angiosarcoma
Lung	LungNeoplasm Adenocarcinoma of the lung Non-small cell lung cancer Lung adenocarcinoma Small cell lung cancer Lung carcinoid Adenosquamous carcinoma of the lung Large cell neuroendocrine carcinoma Salivary gland-type lung carcinoma Mesothelioma
KidneyCortex KidneyMedulla	KidneyNeoplasm Renal cell carcinoma Transitional cell carcinoma Renal sarcoma
Bladder	BladderTumor Papillary carcinoma Flat carcinomas Adenocarcinoma of the bladder Sarcoma
Stomach	StomachNeoplasm Adenocarcinomas of the stomach Gastric carcinoid Carcinoid tumor
Uterus	UterineNeoplasm Uterine cancer Endometrial cancer Uterine sarcoma Serous adenocarcinoma Adenosquamous carcinoma Uterine carcinosarcoma
Testis	TesticularNeoplasm Seminoma Non-seminoma spermatocytic seminoma Recurrent testicular cancer
Pancreas	PancreaticNeoplasm Adenocarcinoma Endocrine tumor Exocrine tumor

EsophagusMucosa	EsophagealSquamousCellCarcinoma Esophageal adenocarcinoma Squamous cell carcinoma
EsophagusGastroesophagealJunction	EsophagealAdenocarcinoma

Comment by [Shamsuddin Sultan Khan](#) [04/Aug/21]

Philipp, I mistakenly put **Triple-negative breast cancer, Metastatic breast cancer** in brain tissue. This would be placed in Breast tissue.

Comment by [Shamsuddin Sultan Khan](#) [05/Aug/21]

Looks good, Philipp.

Comment by [Shamsuddin Sultan Khan](#) [10/Aug/21]

New editing is ok

Comment by [Philipp Anokhin](#) [10/Aug/21]

[Shamsuddin Sultan Khan](#), please check all disease names with question marks agains <https://platform.opentargets.org/>

Comment by [Shamsuddin Sultan Khan](#) [17/Aug/21]

Disease names are ok. if you don't find it in the OT. because it is rare and has very little research data.

Comment by [Philipp Anokhin](#) [18/Aug/21]

As we agreed, [Shamsuddin Sultan Khan](#) will search for all diseases with question marks in OT and will either remove them (use strikethrough font) OR provide a different d hits in OT (use red text color).

Comment by [Shamsuddin Sultan Khan](#) [25/Aug/21]

Disease name verification completed.

Comment by [Shamsuddin Sultan Khan](#) [26/Aug/21]

Philipp, we will include the rare diseases although few targets. we need to add a sub-category for those diseases to cover more targets.

Comment by [Philipp Anokhin](#) [27/Aug/21]

[Dhaval Patel](#), I'm done with the disease list, below is the latest list (data files copied to S3 and Jenkins pipeline re-configured). Please implement in portal.

Tissue	Disease
Prostate	ProstateNeoplasm Adenocarcinoma UrothelialCarcinoma ProstateNeuroendocrineNeoplasm SmallCellCarcinoma MetastaticProstateCancer
Pituitary	PituitaryTumor
BrainCerebellum	Glioma GlioblastomaMultiforme Meningioma Craniopharyngioma Astrocytoma Ependymoma Oligodendroglioma GerminomatousGermCellTumor Medulloblastoma
Breast	BreastNeoplasm Triple-negative breast cancer InvasiveBreastDuctalCarcinoma InvasiveLobularCarcinoma InflammatoryBreastCarcinoma Angiosarcoma PhyllodesTumor
ColonSigmoid ColonTransverse SmallIntestine	IntestinalNeoplasm ColorectalAdenocarcinoma CarcinoidTumor GastrointestinalStromalTumor Leiomyosarcoma
SkinSun	Melanoma BasalCellCarcinoma SkinSquamousCellCarcinoma MerkelCellSkinCancer KaposiSarcoma Angiosarcoma
LymphNodes	FollicularLymphoma
MuscleSkeletal	Rhabdomyosarcoma
Liver	LiverNeoplasm HepatocellularCarcinoma

	Fibrolamellar Cholangiocarcinoma Angiosarcoma
Lung	LungNeoplasm LungAdenocarcinoma NonSmallCellLungCarcinoma SmallCellLungCarcinoma LungCarcinoidTumor AdenosquamousLungCarcinoma SquamousCellLungCarcinoma LargeCellNeuroendocrineCarcinoma AdenoidCysticCarcinoma MucoepidermoidCarcinoma Mesothelioma
KidneyCortex KidneyMedulla	KidneyNeoplasm RenalCellCarcinoma TransitionalCellCarcinoma KidneySarcoma
Bladder	BladderTumor Papillary carcinoma BladderAdenocarcinoma
Stomach	StomachNeoplasm GastricAdenocarcinoma GastricNonCardiaCarcinoma CarcinoidTumor
Uterus	UterineNeoplasm UterineCancer Endometrial cancer UterineSarcoma SerousAdenocarcinoma AdenosquamousCarcinoma UterineCarcinosarcoma
Testis	TesticularNeoplasm Seminoma TesticularNonSeminomatousGermCellTumor
Pancreas	PancreaticNeoplasm PancreaticAdenocarcinoma PancreaticNeuroendocrineTumor MalignantExocrinePancreasNeoplasm
EsophagusMucosa	EsophagealSquamousCellCarcinoma EsophagealAdenocarcinoma
EsophagusGastroesophagealJunction	EsophagealAdenocarcinoma

Comment by [Stuart Ross](#) [07/Sep/21]

Dev complete, now in testing

Comment by [Dhaval Patel](#) [14/Sep/21]

I added all diseases and tissue in the live database but I need help from [Shamsuddin Sultan Khan](#) to find the exact code of the disease.

Comment by [Stuart Ross](#) [14/Sep/21]

Dhaval and Shams to update/map spreadsheet - https://www.icd10data.com/ICD10CM/Codes/Changes/New_Codes/1?year=2021

Comment by [Shamsuddin Sultan Khan](#) [26/Sep/21]

Dhaval, please arrange a meeting with me to complete this disease code work.

Comment by [Dhaval Patel](#) [27/Sep/21]

Today will connect sharp 5PM

Comment by [Dhaval Patel](#) [28/Sep/21]

[Philipp Anokhin](#) Please provide mapping with EFO

Comment by [Shamsuddin Sultan Khan](#) [02/Oct/21]

Please use Malacard disease code instead Open target code. Open target has very limited info for disease diagnosis. We need biological details for diagnosis purposes an

Comment by [Philipp Anokhin](#) [02/Oct/21]

We agreed on EFO codes (used also by GWAS and NIH) for now. Internally pipeline already uses EFO. If we need to map to additional ontologies, we can do it at any time

Comment by [Shamsuddin Sultan Khan](#) [03/Oct/21]

I think I already discussed the rationale of using the Malacard disease code.

If we like EFO code, then just map it with the diseases plz. But we should not put it with the patient accession number.

We are using a code from Malacard to research the proper diagnostic cases, disease mechanisms & pharmacology, not the purpose to follow the NIH or GWAS etc. Using is a different purpose.

I think we haven't agreed to alter the patient accession number. We don't necessarily need to change the accession number format.

Many R&D tasks are pending and I think we might waste time on an established and settled thing.

Comment by [Philipp Anokhin](#) [03/Oct/21]

We have agreed to change a unique patient identifier to a number and change data architecture to reflect that. We have to do it to accommodate multiple diseases/analyses: patient number on the report is a different story and is unrelated to the discussion here - EFO codes and new patient ID format is for internal use only. You can create a separate MalaCard codes or comment on the existing PES report issue.

Comment by [Shamsuddin Sultan Khan](#) [03/Oct/21]

Dhaval needs to explain all of the rationales too as we have changed this three times before. Kevin & Carlos need to be On. Need to be simple.

Comment by [Philipp Anokhin](#) [29/Oct/21]

[Dhaval Patel](#), I've finished mapping diseases to EFO codes - please implement in the portal.

Comment by [Philipp Anokhin](#) [19/Jan/22]

[Dhaval Patel](#), please add "Liposarcoma" to the list of diseases in portal.

Comment by [Philipp Anokhin](#) [20/Jan/22]

[Shamsuddin Sultan Khan](#), please find relevant cancer disease names corresponding to new tissues in the table above (with empty lines) and find their EFO codes on [https](#) Then assign to [Dhaval Patel](#) for changes to portal.

Comment by [Philipp Anokhin](#) [09/May/22]

Update the disease data in PES pipeline.

Comment by [Philipp Anokhin](#) [28/Jun/22]

[Dhaval Patel](#), please update the disease data in portal as soon as possible. This is a large update - check the table in description. I've already updated PES pipeline and up

Comment by [Dhaval Patel](#) [18/Jul/22]

[Philipp Anokhin](#) , I uploaded Disease and tissues.

Comment by [Philipp Anokhin](#) [18/Jul/22]

[Shamsuddin Sultan Khan](#), can you please check the QA portal and try running it for the next patient?

Comment by [Shamsuddin Sultan Khan](#) [19/Jul/22]

Ok Philipp.

[Dhaval Patel](#) Actually, Dhaval needs to show me the tissue selection process at the pipeline run?

Comment by [Philipp Anokhin](#) [14/Sep/22]

Adding Osteosarcoma

Comment by [Philipp Anokhin](#) [14/Sep/22]

[Dhaval Patel](#), please add Osteosarcoma to the disease list in the portal.

Comment by [Stuart Ross](#) [29/Sep/22]

[Shamsuddin Sultan Khan](#) [Dhaval Patel](#) [Philipp Anokhin](#) Data Team, This needs done ASAP! The current list is incomplete, I could not find Burkitts Lymphoma in the Portal c

Comment by [Shamsuddin Sultan Khan](#) [29/Sep/22]

[Philipp Anokhin](#) why do we need Burkitts Lymphoma now? we do not have any patients with this type lymphoma yet. It is not on our master list so Stuart will not find it.

[Dhaval Patel](#) if it is missing. let me know. I will find corresponding tissue

Comment by [Philipp Anokhin](#) [29/Sep/22]

[Stuart Ross](#), why don't you let us know why do you need Burkitts Lymphoma? I'm sure this list doesn't include all possible cancers as that would mean downloading the entire usually add missing diseases when we get a patient that needs it.

Comment by [Philipp Anokhin](#) [26/Oct/22]

[Dhaval Patel](#), please add LeptomeningealMetastasis and BreastDuctalCarcinomaInsitu diseases to portal.

Comment by [Philipp Anokhin](#) [07/Nov/22]

[Dhaval Patel](#), please add MalignantTesticularGermCellTumor disease (in Testis tissue), then **resolve** (not close) this issue.

Comment by [Philipp Anokhin](#) [17/Nov/22]

[Dhaval Patel](#), please add ProstateAdenocarcinoma to Prostate tissue and remove the generic Adenocarcinoma

Comment by [Philipp Anokhin](#) [22/Nov/22]

[Dhaval Patel](#), please add SquamousCellProstateCarcinoma to Prostate tissue