

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

Created: 03/Feb/21 Updated: 13/Mar/23 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 |
|--------------|--------------|---------------------------|---------------------------|

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 |
| Lymphocytes missing | 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 |
| Testis | UterineCarcinosarcoma | EFO_1000613 |
| | TesticularNeoplasm | EFO_0004281 |
| | Seminoma | MONDO_0003001 |
| | TesticularNonSeminomatousGermCellTumor | EFO_1000570 |
| Pancreas | MalignantTesticularGermCellTumor | MONDO_0003510 |
| | PancreaticNeoplasm | EFO_0003860 |
| | PancreaticAdenocarcinoma | EFO_1000044 |
| | PancreaticNeuroendocrineTumor | EFO_1000045 |
| EsophagusMucosa | MalignantExocrinePancreasNeoplasm | MONDO_0002116 |
| | EsophagealSquamousCellCarcinoma | EFO_0005922 |
| EsophagusGastroesophagealJunction | EsophagealAdenocarcinoma | EFO_0000478 |
| | | EFO_0000478 |

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-'
'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_read:
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-Esop:
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-Fallop:
'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-Hea:
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-Heart -
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_
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/analyse: 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 sepa of 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 ent 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