phenopackets-to-fhir

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CHAPTER

ONE

INTRODUCTION

phenopackets-to-fhir is a service to convert derived phenopackets objects to FHIR resources. The service is a part of the ongoing work for CHORD Metadata service. The phenopackets schemas are project-specific adaptations of Phenopackets schema standard.

The package contains:

- json schemas for Phenopacket and its elements derived/based on Phenopackets schema standard.
- functions to transform phenopackets objects to FHIR resources.
- mappings derived from Phenopackets on FHIR Implementation Guide.

The conversion uses Python SMART on FHIR client that provides validation of generated FHIR resources.

CHAPTER

TWO

CONVERTERS

`converters.resources.`**`age_to_fhir`**(*obj*, *mapping*, *field*)

Generic function to convert Phenopackets Age or AgeRange to FHIR Age.

Parameters

- **`obj`** – object to which field Age or AgeRange belongs to
- **`mapping`** – mapping from PHENOPACKETS_ON_FHIR
- **`field`** – name of the field that stores age

Returns age extension object

`converters.resources.`**`biosample_to_fhir`**(*obj*)

Converts Biosample to FHIR Specimen.

Parameters **`obj`** – Biosample json

Returns FHIR Specimen json

`converters.resources.`**`check_disease_onset`**(*disease*)

Phenopackets schema allows age to be represented by ISO8601 string, whereis Pheno-FHIR guide requires it to be represented by CodeableConcept. This function checks how age is represented in data.

Parameters **`disease`** – disease json object

Returns True/False

`converters.resources.`**`codeable_concepts_fields`**(*field_list*, *profile*, *obj*)

Converts a list of fields to FHIR CodeableConcepts and returns a list of extensions.

Parameters

- **`field_list`** – fields to convert to codeable concepts
- **`profile`** – name of an object in phenopackets mappings
- **`obj`** – object to which fields belong to

Returns list of extensions

`converters.resources.`**`disease_to_fhir`**(*obj*)

Converts Disease to FHIR Condition.

Parameters **`obj`** – Disease json

Returns FHIR Condition json

`converters.resources.`**`fhir_codeable_concept`**(*obj*)

Generic function to convert object to FHIR CodeableConcept.

`converters.resources.`**`fhir_coding_util`**(*obj*)
Genenric function to convert object to FHIR Coding.

`converters.resources.`**`gene_to_fhir`**(*obj*)
Convert Gene to Observation.component:gene property. GA4GH Phenopackets Implementation Guide provides a link to Genomics Reporting Implementation Guide (STU1) mapping http://hl7.org/fhir/uv/genomics-reporting/StructureDefinition/region-studied .

Parameters **`obj`** – gene json

Returns Observation.component:gene json

`converters.resources.`**`hts_file_to_fhir`**(*obj*)
Converts HTS file to FHIR DocumentReference.

Parameters **`obj`** – HTS file json

Returns FHIR DocumentReference json

`converters.resources.`**`individual_to_fhir`**(*obj*)
Converts Individual to FHIR Patient.

Parameters **`obj`** – Individual json

Returns FHIR Patient json

`converters.resources.`**`phenopacket_to_fhir`**(*obj*)
Converts Phenopacket to FHIR Composition.

Parameters **`obj`** – Phenopacket json

Returns FHIR Composition json

`converters.resources.`**`phenotypic_feature_to_fhir`**(*obj*)
Converts Phenotypic feature to FHIR Observation.

Parameters **`obj`** – PhenotypicFeature json

Returns FHIR Observation json

`converters.resources.`**`procedure_to_fhir`**(*obj*)
Converts Procedure to FHIR Specimen collection.

`converters.resources.`**`variant_to_fhir`**(*obj*)
Variant corresponds to Observation.component:variant. GA4GH Phenopackets Implementation Guide provides a link to Genomics Reporting Implementation Guide (STU1) mapping http://hl7.org/fhir/uv/genomics-reporting/STU1/variant.html.

Parameters **`obj`** – Variant json

Returns Observation.component:variant json

CHAPTER

THREE

VALIDATION

`converters.validate.`**`validate_schema`**(*schema*, *data*)

Validates the instance against corresponding json schema.

Parameters

- **`schema`** – the json schema to validate with
- **`data`** – the instance to validate

Returns True if the instance is valid, False if not

Raises *ValidationError* if the instance is invalid

CHAPTER

FOUR

MAPPINGS

4.1 HL7 Genomics Mappings

```
# All fixed values provided by Phenopackets on FHIR Mapping guide
# Mappings from http://build.fhir.org/ig/HL7/genomics-reporting/index.html

HL7_GENOMICS_MAPPING = {
    "gene": {
        "url": "http://hl7.org/fhir/uv/genomics-reporting/StructureDefinition/region-
↪studied",
        "geneStudied_Code": {
            "system": "https://loinc.org",
            "id": "48018-6",
            "label": "Gene studied [ID]"
        },
        "geneStudiedValue": {
            "system": "https://www.genenames.org/"
        }
    },
    "variant": {
        "url": "http://hl7.org/fhir/uv/genomics-reporting/StructureDefinition/variant
↪",
        "variantLengthCode": {
            "system": "https://loinc.org",
            "id": "81300-6",
            "label": "Structural variant [Length]"
        }
    }
}
```

4.2 Phenopackets on FHIR Mappings

```
# All fixed values provided by Phenopackets on FHIR Mapping guide

PHENOPACKETS_ON_FHIR_MAPPING = {
    "individual": {
        "title": "Individual",
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/Individual",
        "age": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/individual-age
↪",
        "karyotypicSex": {
```

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```
            "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/individual-
↪karyotypic-sex",
            "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/karyotypic-sex"
        },
        "taxonomy": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪individual-taxonomy"
    },
    "phenopacket": {
        "title": "Phenopacket",
        "code": {
            "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/document-type",
            "code": "phenopacket"
        },
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/Phenopacket",
        "phenotypicFeatures": {
            "title": "Phenotypic Features",
            "code": {
                "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/section-type
↪",
                "version": "0.1.0",
                "code": "phenotypic-features",
                "display": "Phenotypic Features"
            }
        },
        "biosamples": {
                "title": "Biosamples",
                "code": {
                    "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/section-
↪type",
                    "version": "0.1.0",
                    "code": "biosamples",
                    "display": "Biosamples"
                }
            },
        "variants": {
            "title": "Variants",
            "code": {
                "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/section-type
↪",
                "version": "0.1.0",
                "code": "variants",
                "display": "Variants"
            }
        },
        "diseases": {
            "title": "Diseases",
            "code": {
                "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/section-type
↪",
                "version": "0.1.0",
                "code": "diseases",
                "display": "Diseases"
            }
        },
        "htsFiles": {
            "title": "HTS Files",
            "code": {
```

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```
                "system": "http://ga4gh.org/fhir/phenopackets/CodeSystem/section-type
↪",
                "version": "0.1.0",
                "code": "hts-files",
                "display": "HTS Files"
            }
        }
    },
    "biosample": {
        "title": "Biosample",
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/Biosample",
        "individualAgeAtCollection": "http://ga4gh.org/fhir/phenopackets/
↪StructureDefinition/biosample-individual-age-at-collection",
        "histologicalDiagnosis": "http://ga4gh.org/fhir/phenopackets/
↪StructureDefinition/biosample-histological-diagnosis",
        "tumorProgression": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪biosample-tumor-progression",
        "tumorGrade": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪biosample-tumor-grade",
        "diagnosticMarkers": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪biosample-diagnostic-markers",
        "isControlSample": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪biosample-control"
    },
    "phenotypicFeature": {
        "title": "Phenotypic Feature",
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪PhenotypicFeature",
        "severity": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪phenotypic-feature-severity",
        "modifier": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪phenotypic-feature-modifier",
        "onset": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/phenotypic-
↪feature-onset",
        "evidence": {
            "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/evidence",
            # fixed value
            "evidenceCode": "evidenceCode"
        }
    },
    "externalReference": {
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/external-
↪reference",
        # fixed values
        "idUrl": "id",
        "descriptionUrl": "description"
    },
    "htsFile": {
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/HtsFile",
        "genomeAssembly": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪htsfile-genome-assembly",
        # fixed value
        "status": "current",
        "htsFormat": "http://ga4gh.org/fhir/phenopackets/CodeSystem/hts-format"
    },
    "disease": {
        "url": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/Disease",
```

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```
        "onset": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/disease-onset
↪",
        "disease_stage": "http://ga4gh.org/fhir/phenopackets/StructureDefinition/
↪disease-tumor-stage",
    }
}
```

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FIVE

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