

**Title:** A formalization of one of the main claims of “Overlap of vitamin A and vitamin D target genes with CAKUT-related processes” by Ozisik et al. 2021

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<http://purl.org/np/RAyg4UqIVovBGia-hk4qEuRzOg14fcOIYAclC6YQGQaVYU>

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## **Abstract:**

In a previous paper that we co-authored (Ozisik et al. 2021) we showed that in some cases Vitamin A and Vitamin D receptor binding sites are present in genes associated with Congenital Anomalies of the Kidney and Urinary Tract (CAKUT). From that finding we derived the statement that sometimes Vitamin A targets are the same as genes associated with CAKUT. We present here a formalization of that claim, stating that all things of class “genes associated with CAKUT” sometimes have a relation of type “is same as” to a thing of class “targets of vitamin A”.

## **1. Introduction**

We present here a formalization of the main scientific claim from Ozisik et al. [1] by using a semantic template called the super-pattern [2].

## **2. Formalization**

Our formalization looks as follows:

CONTEXT-CLASS (“in the context of all ...”): [\(universal context\)](#)

SUBJECT-CLASS (“things of type ...”): [genes associated with CAKUT](#)

QUALIFIER: [sometimes](#)

RELATION-TYPE (“have a relation of [is same as](#) type...”):

OBJECT-CLASS (“to things of type...”): [targets of vitamin A](#)

In the context class we use the class “universal context” from SuperPattern ontology. In the subject class, we use the class “genes associated with CAKUT” (Q109406970) from Wikidata. In the object class we use the class “targets of vitamin A” (Q109406949) from Wikidata.

### 3. RDF Code

This is our formalization as a nanopublication in TriG format:

```
@prefix this: <http://purl.org/np/RAyg4UgIVovBGia-hk4qEuRzOq14fcOlYAclC6YGQaVYU> .
@prefix sub: <http://purl.org/np/RAyg4UgIVovBGia-hk4qEuRzOq14fcOlYAclC6YGQaVYU#> .
@prefix np: <http://www.nanopub.org/nschema#> .
@prefix dct: <http://purl.org/dc/terms/> .
@prefix nt: <https://w3id.org/np/o/ntemplate/> .
@prefix npx: <http://purl.org/nanopub/x/> .
@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .
@prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#> .
@prefix orcid: <https://orcid.org/> .
@prefix prov: <http://www.w3.org/ns/prov#> .
@prefix sp: <<https://w3id.org/linkflows/superpattern/terms/>> .

sub:Head {
  this: np:hasAssertion sub:assertion ;
  np:hasProvenance sub:provenance ;
  np:hasPublicationInfo sub:pubinfo ;
  a np:Nanopublication .
}
sub:assertion {
  sub:spi a <https://w3id.org/linkflows/superpattern/terms/SuperPatternInstance> ;
  rdfs:label "Sometimes Vitamin A targets are the same as genes associated with CAKUT" ;
  sp:hasContextClass sp:UniversalContext ;
  sp:hasSubjectClass <https://www.wikidata.org/wiki/Q109406970> ;
  sp:hasQualifier sp:sometimesQualifier ;
  sp:hasRelation sp:isSameAs ;
  sp:hasObjectClass <https://www.wikidata.org/wiki/Q109406949> .
}
sub:provenance {
  sub:activity a sp:FormalizationActivity ;
  prov:used <https://doi.org/10.12688/f1000research.51018.1> ;
  prov:wasAssociatedWith orcid:0000-0002-5301-3142 , orcid:0000-0002-7770-620X .
  sub:assertion prov:wasGeneratedBy sub:activity .
}
sub:pubinfo {
  sub:sig npx:hasAlgorithm "RSA" ;
  npx:hasPublicKey
    "MIGfMA0GCSqGSIb3DQEBQUAA4GNADCBiQKBgQCCYtnHL3Zu9ExrWA28zHnRA9JKmj8V9awChjn+7oBn9p6wdLx6lJ5mD/1WK9H8NdxSk/fdoaEJAin2WLiWs6qBjdsZ
    OLbfq7HSF/GVYoTkuvvjnf2rozS08mI+XwEXMsa7XVa+4bz8jauPyp/eEFbcIHEWtstiuQvjvoXKizK5HQIDAQAB" ;
  npx:hasSignature
    "I3KzDEFUZEyP4oMCYAlMchEGcmMpEmFjZLwDF/TtxkXD1yoRj40BopL4n78X+5ldUVsO4ufYlJ8JXe+TFgPXX/Z9mBLE+Ebzhux7pGRG9woM16C5fLTwrJoJL7rcUK6i
    i7moFZ/LsChOkdtC0DrZuie+BecFjvt6xKi693gPih4=" ;
  npx:hasSignatureTarget this: .
  this: dct:created "2021-11-15T09:46:19.034+01:00"^^xsd:dateTime ;
  dct:creator orcid:0000-0002-7770-620X ;
```

```

npx:introduces sub:spi ;
npx:supersedes <http://purl.org/np/RAokVMmiZSbRh01diNeJLum4p13kUd-NZjGFuVtxVz4Bs> ;
<https://w3id.org/linkflows/reviews/isUpdateOf> <http://purl.org/np/RAsdV8EQ1qn_lXOrgoG7mPaF1JXdFLzt2iYy4eMhMMuM4> ;
nt:wasCreatedFromProvenanceTemplate <http://purl.org/np/RAE1wniOy0y039P1K9QkQ-wqbC3q-R2nXraP5huu8W39k> ;
nt:wasCreatedFromPubinfoTemplate <http://purl.org/np/RA2vCBXZf-icEcVRGhulJXugTGxpsV5yVr9yqCI1bQh4A> ,
<http://purl.org/np/RAA2MfqdBczmz9yVWjKLXNbyfBNcwsMmOqcNUxkk1maIM> ,
<http://purl.org/np/RAjpbMlw3owYhJUBo3DtsuD1XsNAJ8cnGeWAutDVjuAuI> ;
nt:wasCreatedFromTemplate <http://purl.org/np/RAv68imZrEjfc2rnEg1hzoBqEVc0cQMtp9_1Za0BxNM4> .
}

```

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## References

- [1] Ozisik O, Ehrhart F, Evelo CT et al. Overlap of vitamin A and vitamin D target genes with CAKUT-related processes [version 1; peer review: 2 approved with reservations]. F1000Research 2021, 395(10), doi: 10.12688/f1000research.51018.1.
- [2] Bucur, C.I., Kuhn, T., Ceolin, D., Ossenbruggen, J. van. Expressing high-level scientific claims with formal semantics. In: Proceedings of the 11th Knowledge Capture Conference 2021. doi: 10.1145/3460210.3493561.