LEIDEN UNIVERSITY MEDICAL CENTER

National Variant Database Proposal

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Introduction

Sources for the database:

- Full genome sequencing.
- Exome sequencing.

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- Annotation of variants called by our pipeline.
  - In how many percent of the cases have we seen this variant.
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  - Annotation of other sources.
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- Triplet studies.
  - Process three samples and query the database for de-novo mutations.

Requirements

The local database currently stores the following data:

- Description of the variant.
  - Chromosome.
  - Start / End position.
  - Reference allele.
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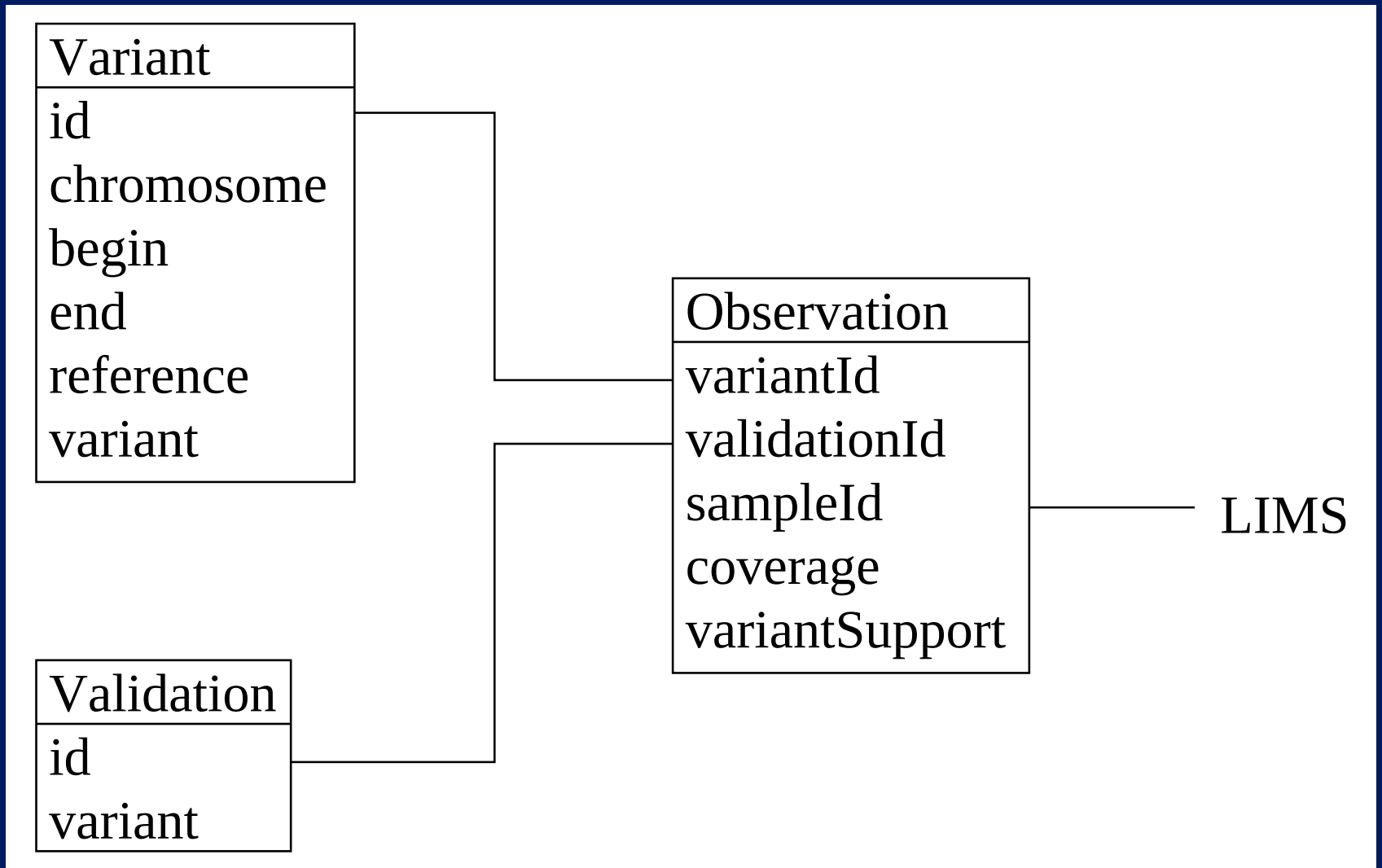
  - ID of an observer.
- Horizontal coverage of each sample.

Layout


Variant
id
chromosome
begin
end
reference
variant
Observation
variantId
validationId
sampleId
coverage
variantSupport
LIMS
Validation
id
variant

Interface: LOVD

Leiden Open Variation Database (LOVD) as an interface.

- Online.
- Gene-centered.
- *Human Genome Variation Society* (HVGS) format.
- Variants are automatically checked (Mutalyzer).
- Variants are described on multiple transcripts (LOVD 3.0 / Mutalyzer).

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To use this interface:

- Variants from the centralised database must be distributed over separate LOVD installations.
  - Known LOVD databases.
  - Currently empty databases for all “Mendelian genes”.
  - Currently non-existent databases for large portions of chromosomes.

`http://www.lovd.nl`

Questions?

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