

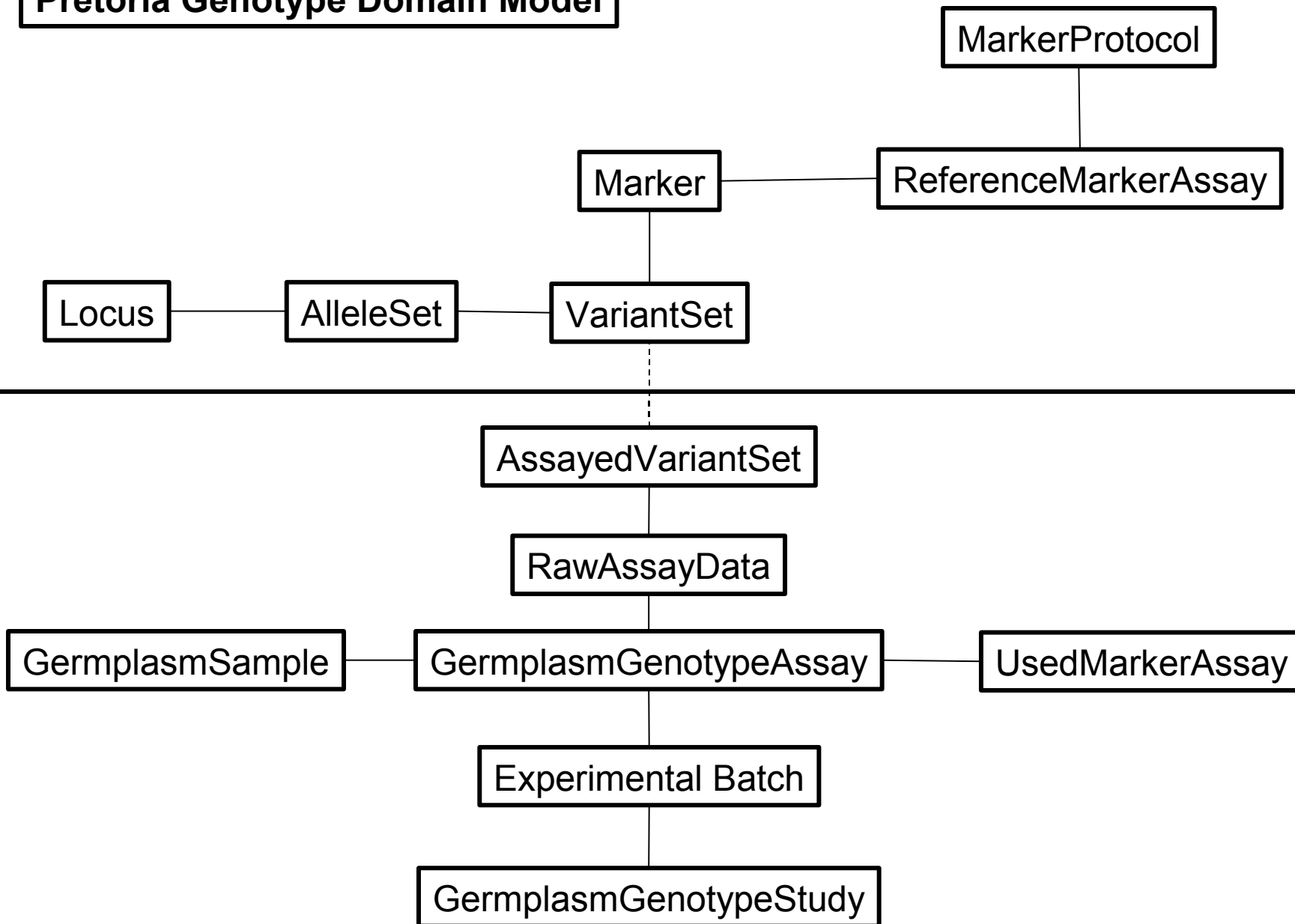
# Genotype Domain Model

Pretoria workshop  
report

# Summary

- The final version of the “Pretoria Model” was validated against several use cases, without major concerns.
- A UML version, additional documentation for the “Pretoria Model” and uses cases is needed. GCPWiki used for documentation and comments.
- Post-Pretoria consensus process is needed.

# Pretoria Genotype Domain Model



# Pretoria model Description

- **AssayedVariantSet:**
  - Validated/curated data that will be used for further analysis. May be referenced to VariantSet.
- **RawAssayData:**
  - Machine or human output. With evidence indication. Can be on different levels, e.g. image, trace files, calls
- **GermplasmGenotypeStudy:**
  - Aggregation of Experimental Batches according to a scientific purpose.
- **Experimental Batch:**
  - GermplasmGenotypeAssays that are performed together, e.g. gel. Can be nested. Captures different levels of experimental errors.
- **GermplasmGenotypeStudy:**
  - Aggregation of GermplasmGenotypeAssays.
- **Used MarkerAssay:**
  - Specific MarkerAssay (from the ReferenceMarkerAssay) that is applied in the GermplasmGenotypeAssay.
- **GermplasmSample:**
  - Biological sample to be assayed. Could be entire plant, plant part, DNA sample.
- **VariantSet:**
  - Set of states in which a Marker is known to exist.
- **Marker:**
  - A landmark used in genotyping.
- **ReferenceMarkerAssay:**
  - MarkerProtocol applied to a specific Marker (specified in Marker)
- **MarkerProtocol:**
  - The experimental conditions can apply to a set of markers. E.g. PCR conditions, Gel type, Sequencer. Does not include person, date.
  - Note: for AFLP marker types the primer pair is part of the MarkerProtocol.

# Use Case

## **AFLP characterization of CGN lettuce collection in the framework of a large EU project.**

- **Marker**
  - a specific location on the gel given a specific AFLP primer combination (a band that is either present or absent)
- **VariantSet**
  - possible states of the marker – usually present or absent, but could also be a quantitative score
- **MarkerProtocol**
  - the published protocol for doing an AFLP analysis (Vos et al.) with some local adjustments the experimental conditions and equipment used (ABI 371 sequencer, type of gel, etc) the method of scoring the gel (visual inspection, reference positions)
- **ReferenceMarkerAssay**
  - combination of Marker and MarkerProtocol (in this case no additional information)
- **UsedMarkerAssay**
  - ReferenceMarkerAssay that was used
- **GermplasmSample**
  - the DNA sample that was used (extracted from a specific plant of a specific CGN accession using a specific DNA extraction protocol, etc.)

# Use Case, continued

- **GermplasmSample**
  - the DNA sample that was used (extracted from a specific plant of a specific CGN accession using a specific DNA extraction protocol, etc.)
- **GermplasmGenotypeStudy**
  - info about the EU project (number, objective, approach, etc)
- **ExperimentalBatch**
  - the composition of the 96 samples on the gel, including info about the standards etc. possibly nested with info about all gels, etc., gel ID (linked to picture of the gel), name of experimenter, identification of the lab, date of experiment
- **GermplasmGenotypeAssay**
  - specific location of a specific marker on a specific lane on a specific gel
- **RawAssayData**
  - call of the band/genotype combination (present or absent / quantitative)
- **AssayedVariantSet**
  - the call of the band/genotype combination after validation, possibly info about the reliability of this call

# Milestones

- Updated UML model on the GCPWiki & GCP model repository (2 weeks)
- Updated “Pretoria model” description on the GCPWiki and reviewed/commented by participants (2 weeks)
- Updated used cases (5) on the GCPWiki (2 weeks)

# CVO's

- No need for CVO's

# Process

- Mailing list for genotype group participants
- Documentation on the GCPWiki
- Consensus process, especially with respect to UML model representing the “consensus model” from the Pretoria workshop (4 weeks)
- Action: T. Metz