



UNIVERSITY OF
LEICESTER

AI-POWERED DSML FOR RARE DISEASES

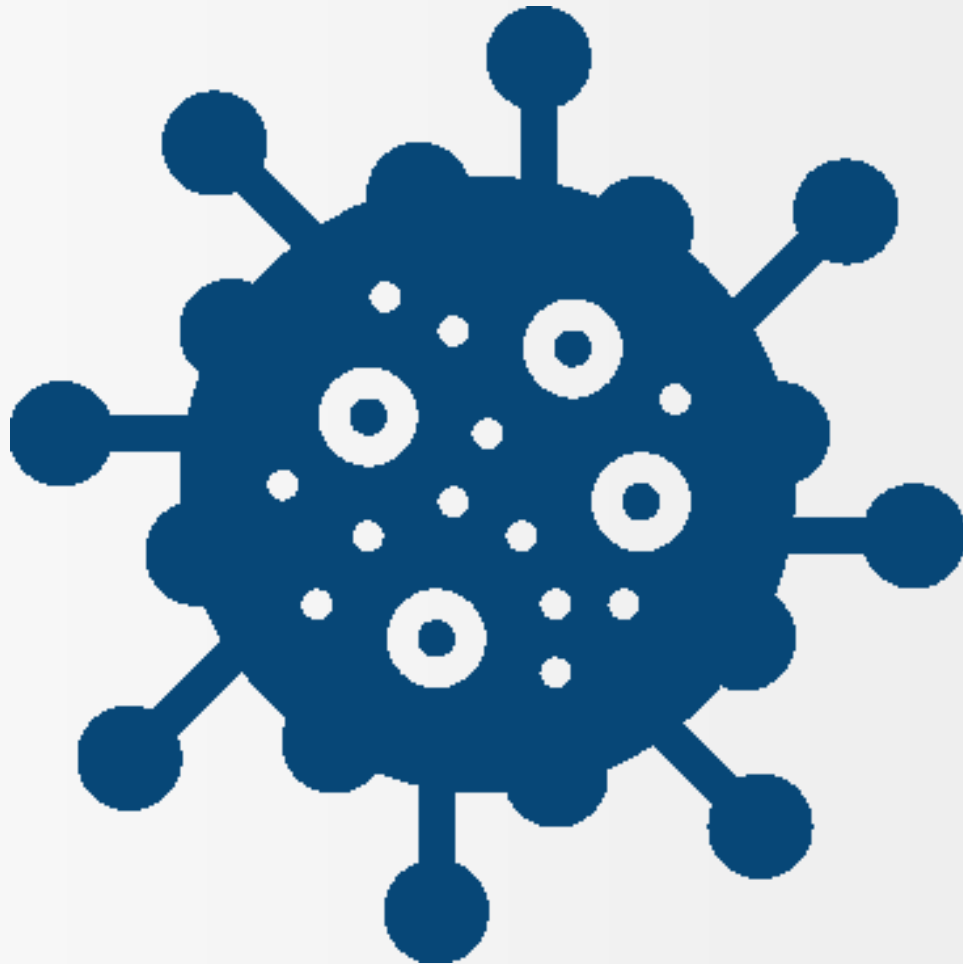
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AGENDA

MOTIVATION

AIM & OBJECTIVES

SYSTEM ARCHITECTURE

CHAIN OF TRANSFORMATION

YAMTL MODEL TRANSFORMATIONS

YAMTL DECLARATIVE RULES

RESULT

CONCLUSION AND FUTURE WORKS

Motivations

- To reduce the complexity encountered by Clinicians in developing query interfaces for data discovery.
- To aid in quicker treatment of patients affected with rare diseases, by empowering medical researchers with a data-adaptive tools for building query UIs.



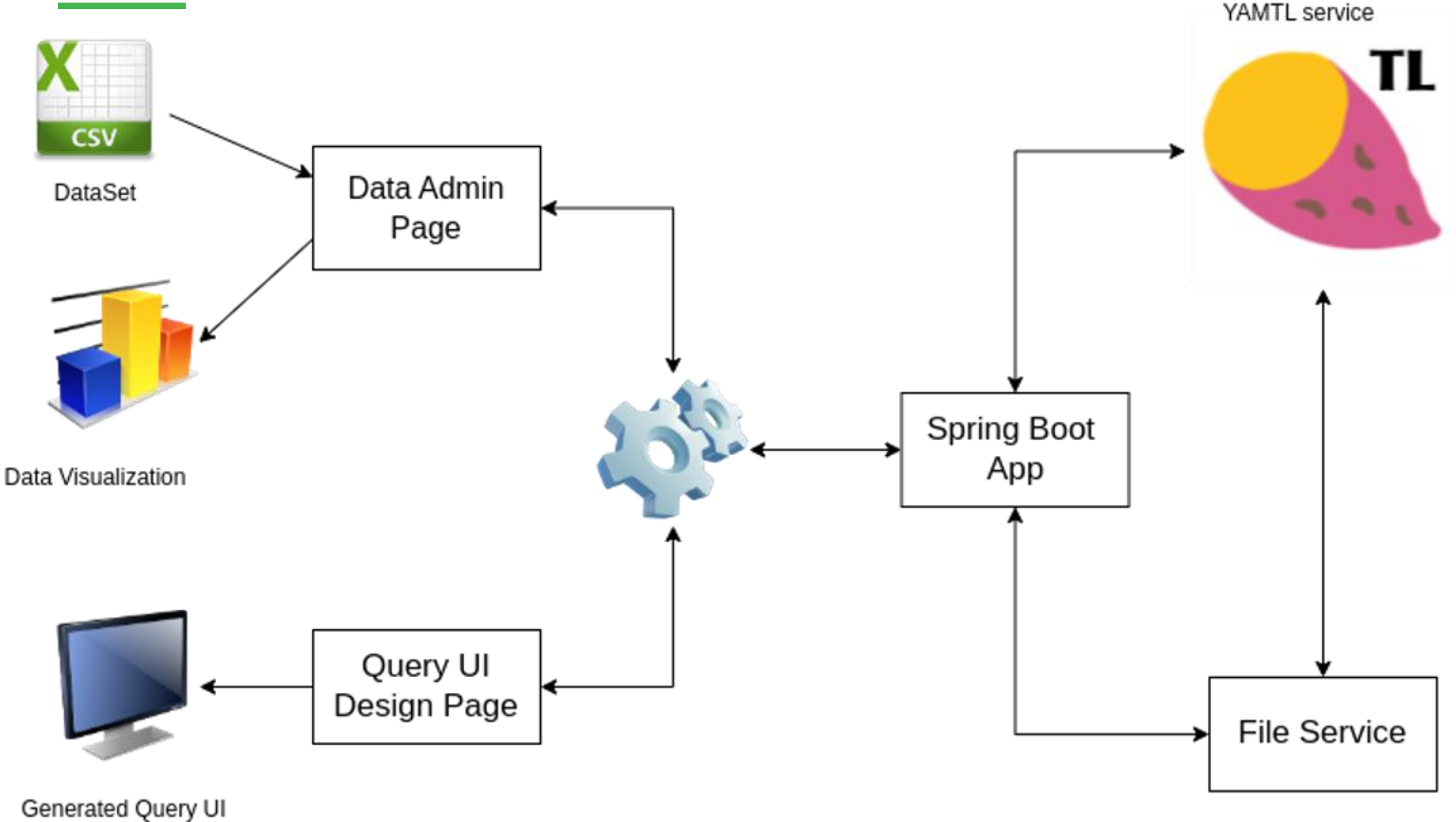
AIM

This project aims to produce a proof-of-concept low code platform for developing optimized query UI's for rare diseases.

OBJECTIVES

- Design of domain-specific languages for analysing datasets, designing UIs, and maintaining them
- Declarative model transformation to obtain query UI models from datasets using YAMTL
- Semantics of UI models in terms of a ReactJS UI (front-end) and propositional logic
- Domain-specific recommendations for building optimized query UIs
 - Learning conceptual UI models from datasets
 - Optimizations in the generated queries

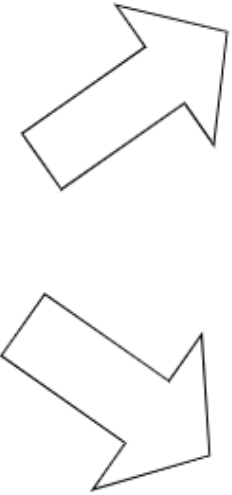
SYSTEM ARCHITECTURE



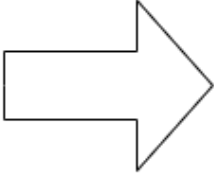
CHAIN OF TRANSFORMATION



DataSet



Description Statistics



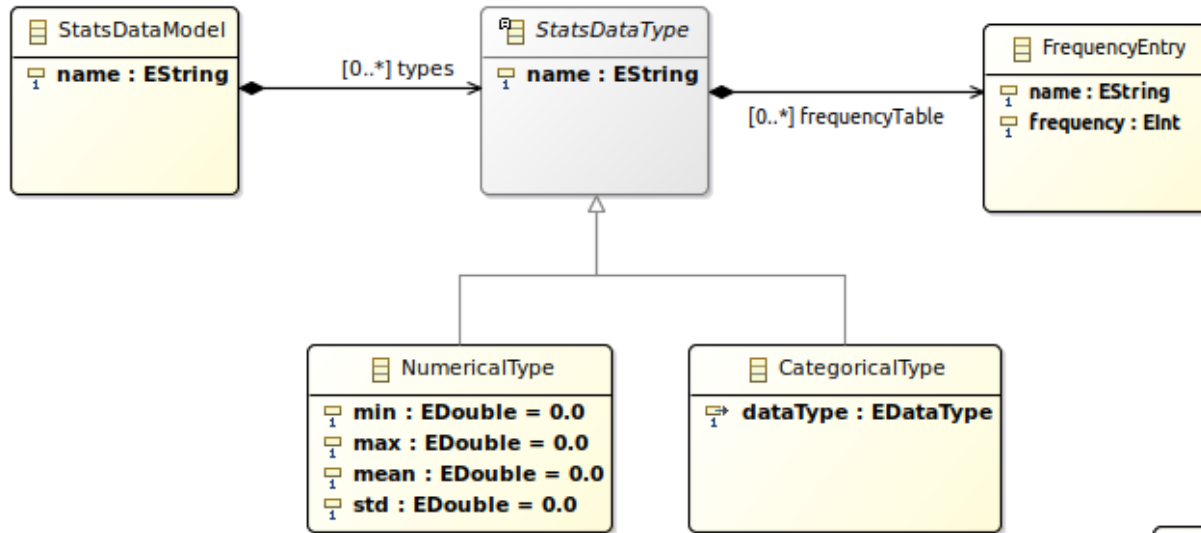
Presentation Model(DSL)



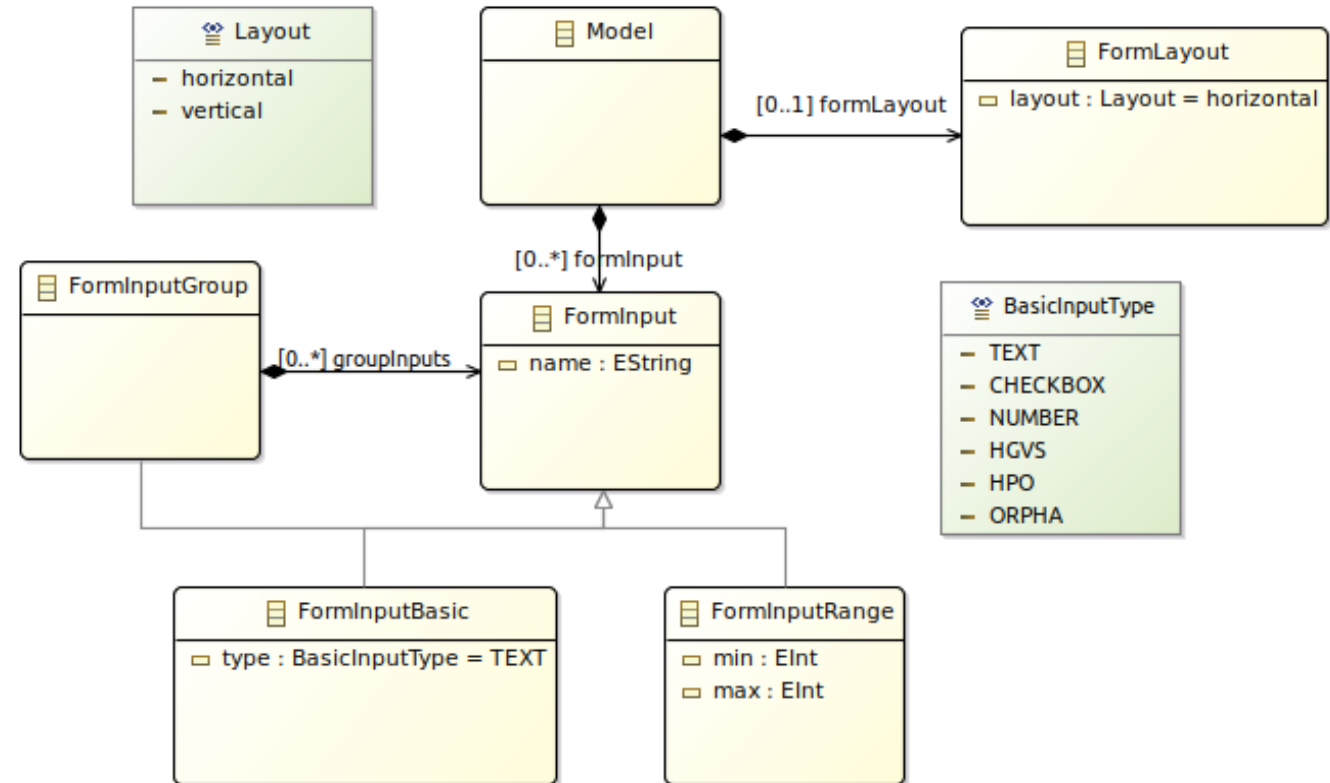
EditProperties

MT YAMTL-(Statistics Model to Presentation Model(DSML))

Source metamodel - Descriptive Statistics Model

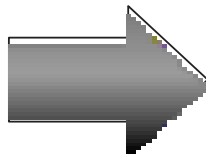
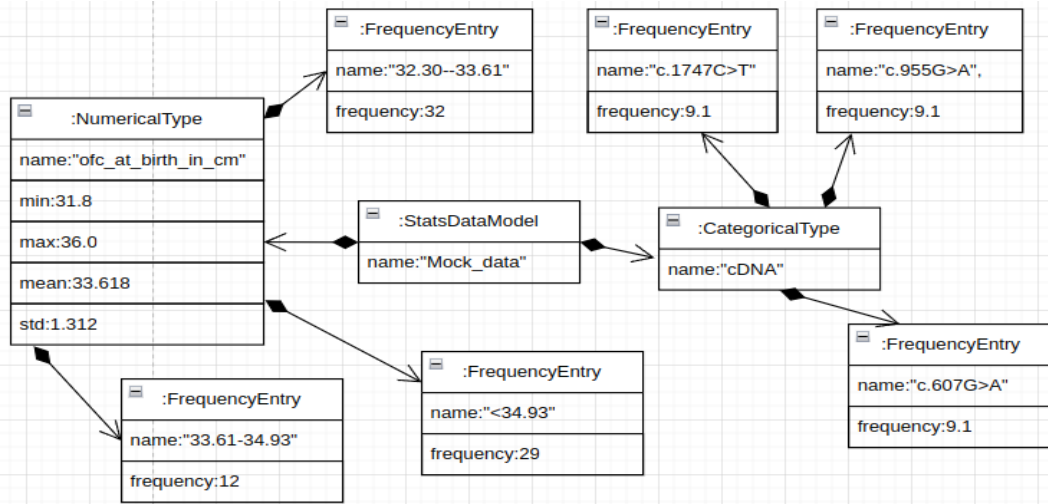


Target metamodel - Presentation Model (VForm)



MT YAMTL-(Statistics Model to Presentation Model(DSML))

Initial source model M_S



DNA SEQUENCE

cDNA
|c.000A>A

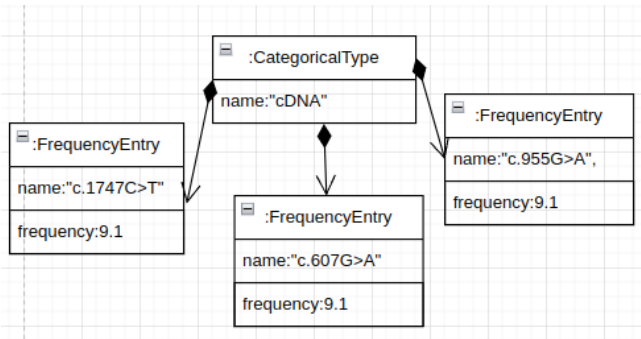
DNA SEQUENCE Term Pairwise Similarity

Minimum Matched Terms

DEMOGRAPHICS

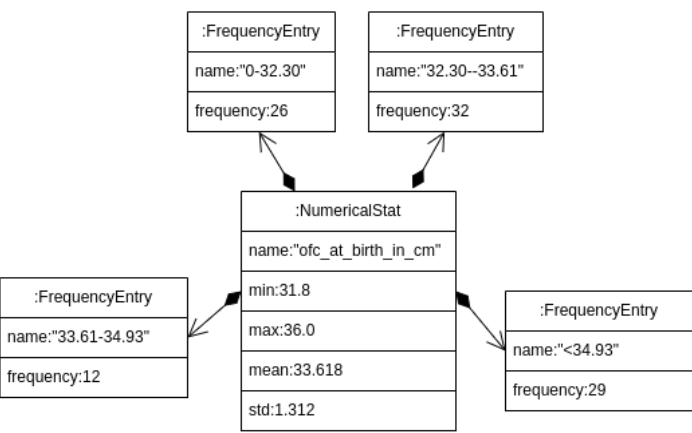
ofc_at_birth_in_cm

YAMTL Rules(Statistics Model to Presentation Model(DSML))



```

rule("hgvs")
  .in("ct", DD.categoricalType).filter[ct.hgvsMatch]
  .out("fib", VF.formInputBasic)[
    val fin = ct.mapFormInput
    fib.name=displayName(ct)
    fib.id = ct.name
    fib.QC = 'true'
    fib.type = "\"hgvs\""
    fin+=fib
  ]
  
```



```

rule('Range')
  .in('nt', DD.numericalType).filter[nt.rangeMatch]
  .out('rg', VF.formInputRange) [
    val fin = nt.mapFormInput
    rg.name = displayName(nt)
    rg.id = nt.name
    rg.QC = 'true'
    rg.min = Math.toIntExact(nt.min)
    rg.max = Math.toIntExact(nt.max)
    fin+=rg
  ]
  
```

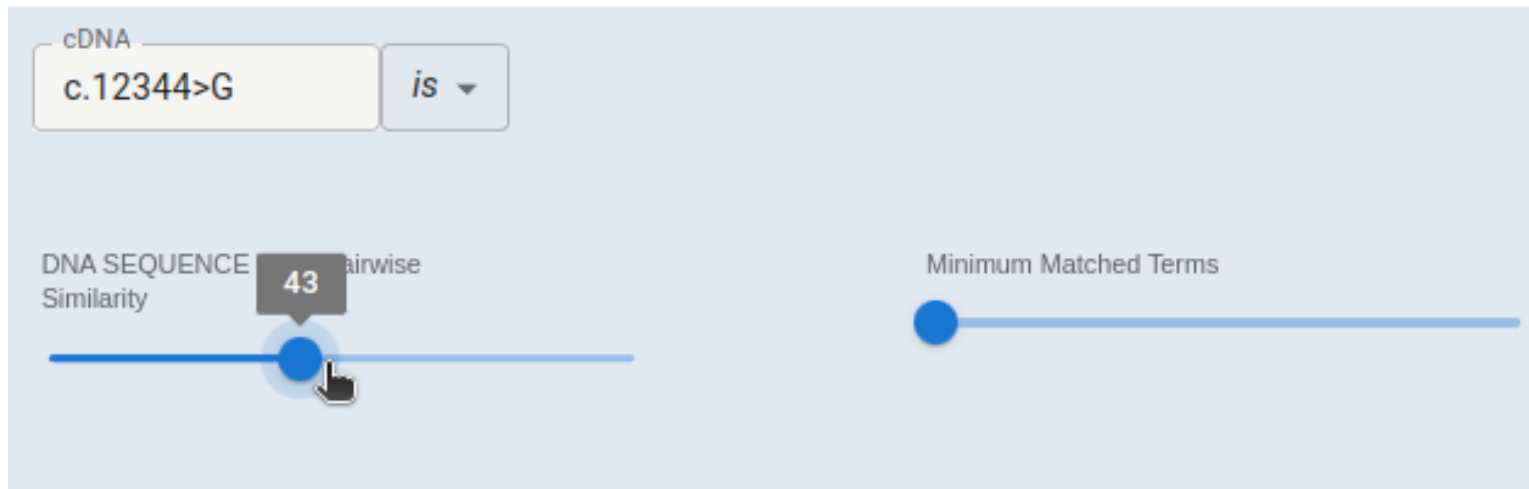
1. GENERATED QUERY VISUAL USER INTERFACE

DNA SEQUENCE

cDNA
c.12344>G *is* ▾

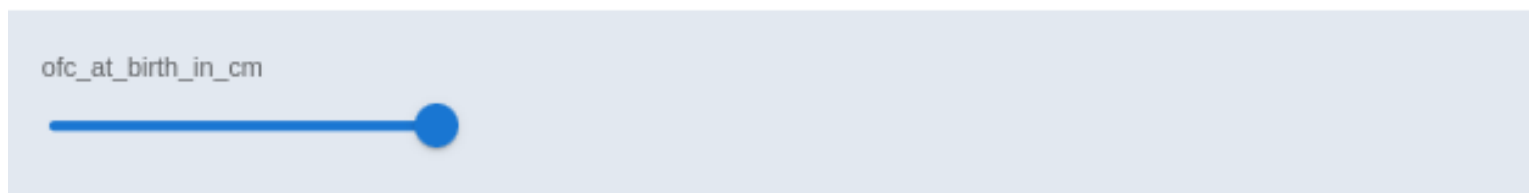
DNA SEQUENCE Similarity **43** pairwise

Minimum Matched Terms



DEMOGRAPHICS


ofc_at_birth_in_cm



```
{  
  "DNA_SEQUENCE": {  
    "cDNA": "IS NOT c.12344>G",  
    "BOOLEAN_LOGIC": "AND",  
    "SIMILARITY": "43% data",  
    "MATCH": "0% data"},  
  
  "DEMOGRAPHICS": {  
    "ofc_at_birth_in_cm": 36,  
    "BOOLEAN_LOGIC": "AND"  
  }  
}
```

1. GENERATED QUERY VISUAL USER INTERFACE

ofc_at_birth_in_cm



id
ofc_at_birth_in_cm

inputName
OFC at Birth

inputType

- none
- range
- select
- number

OFC at Birth

cDNA

c.1234>G

cDNA

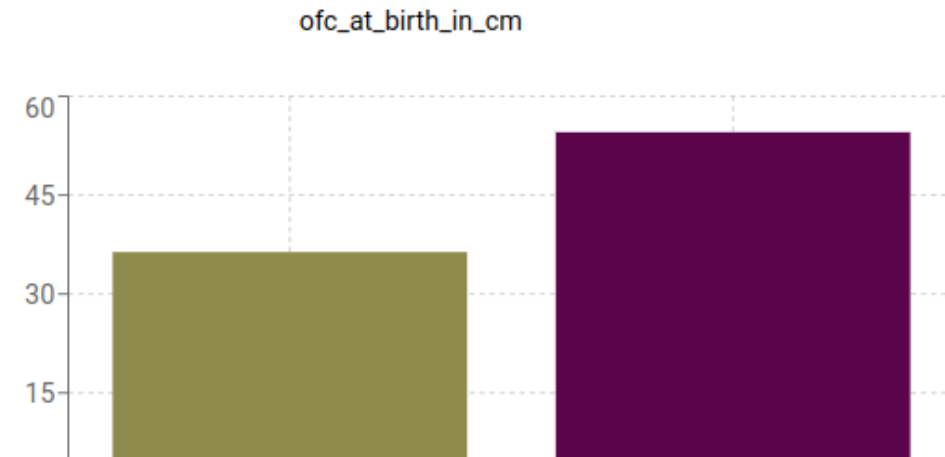
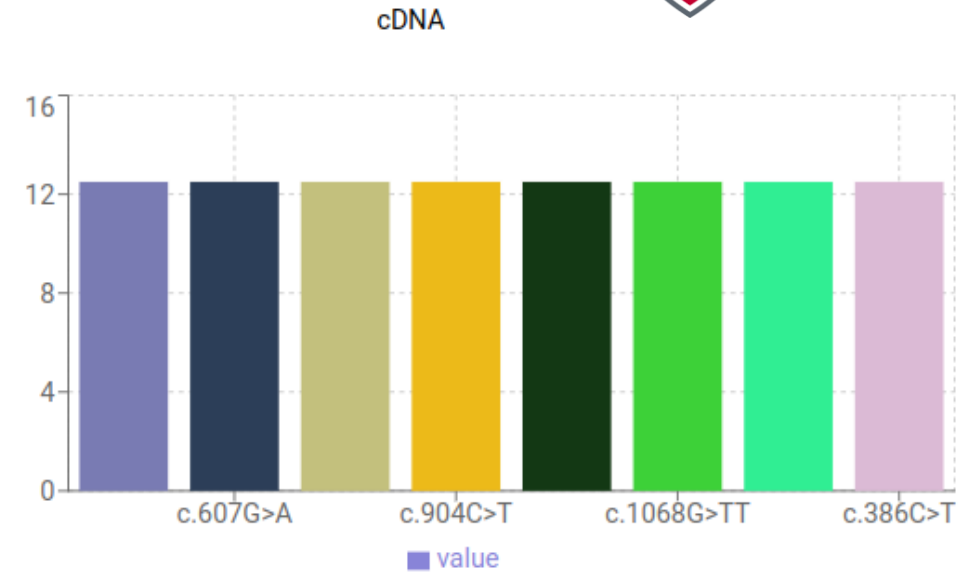
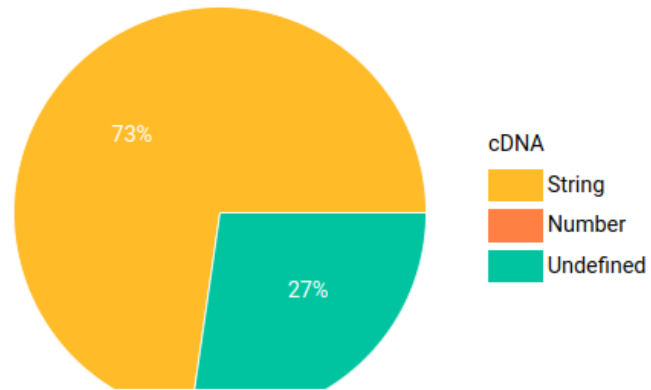
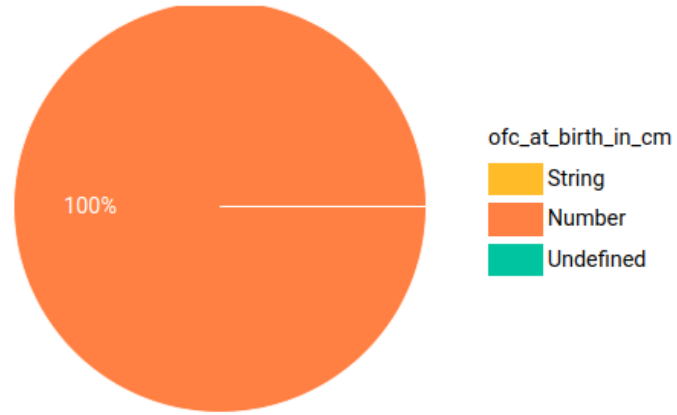
c.5S3>G

please enter a valid HGVS
SEQUENCE

RESULTS



GENERATED VISUALIZATION OF DATASET



Conclusion

The V-Form DSML and inferencing tool helps to abstract away some of the technical challenges faced by medical researchers in data discovery, by automating the creation of query UIs directly from datasets and making provisions for an adaptive approach in customizing the generated UIs. This process has been achieved by eliciting the requirements of the sub domains involved, storing these requirements in models and specifying declarative rules to aid relevant transformations.

Future works

- Integration of Machine Learning Algorithms to efficiently detect genomic data types
- Integration of Query Optimization techniques/heuristics