

# Precision Medicine Knowledge Graph

*By Syed Irtaza Raza  
Software and Biomedical Engineer  
@syedirtazaraza*



# GRAKN.AI

THE KNOWLEDGE GRAPH

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# What Is Precision Medicine?

Precision medicine is an emerging approach for disease **treatment** and **prevention** that takes into account **individual variability** in genes, environment, and lifestyle for each person.



# What Are The Challenges In Precision Medicine?



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# What Are The Challenges In Precision Medicine?

## Integration

Difficult to ingest and integrate  
**multi-format** and **disparate** datasets



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## Integration

Difficult to ingest and integrate  
**multi-format** and **disparate** datasets

## Normalisation

Difficult to maintain **data integrity** of  
**un-structured** complex data



# What Are The Challenges In Precision Medicine?

## Integration

Difficult to ingest and integrate **multi-format** and **disparate** datasets

## Normalisation

Difficult to maintain **data integrity** of **un-structured** complex data

## Discovery

Difficult to **investigate insights** over a magnitude of data in a scalable way



# How Do We Solve These Challenges?

Integration

Normalisation

Discovery



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# How Do We Solve These Challenges?

## Integration

Ingest and integrate complex networks of patient profiles and biological data into one collection – a knowledge base

## Normalisation

## Discovery



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Use automated deductive reasoning to discover and interpret early diagnoses and recommend relevant therapeutic regimes





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Ingest and integrate complex networks of patient profiles and biological data into one knowledge base

## Normalisation

Impose an explicit structure on the data to contextualise the relationships within multi-omics and patient networks

## Discovery

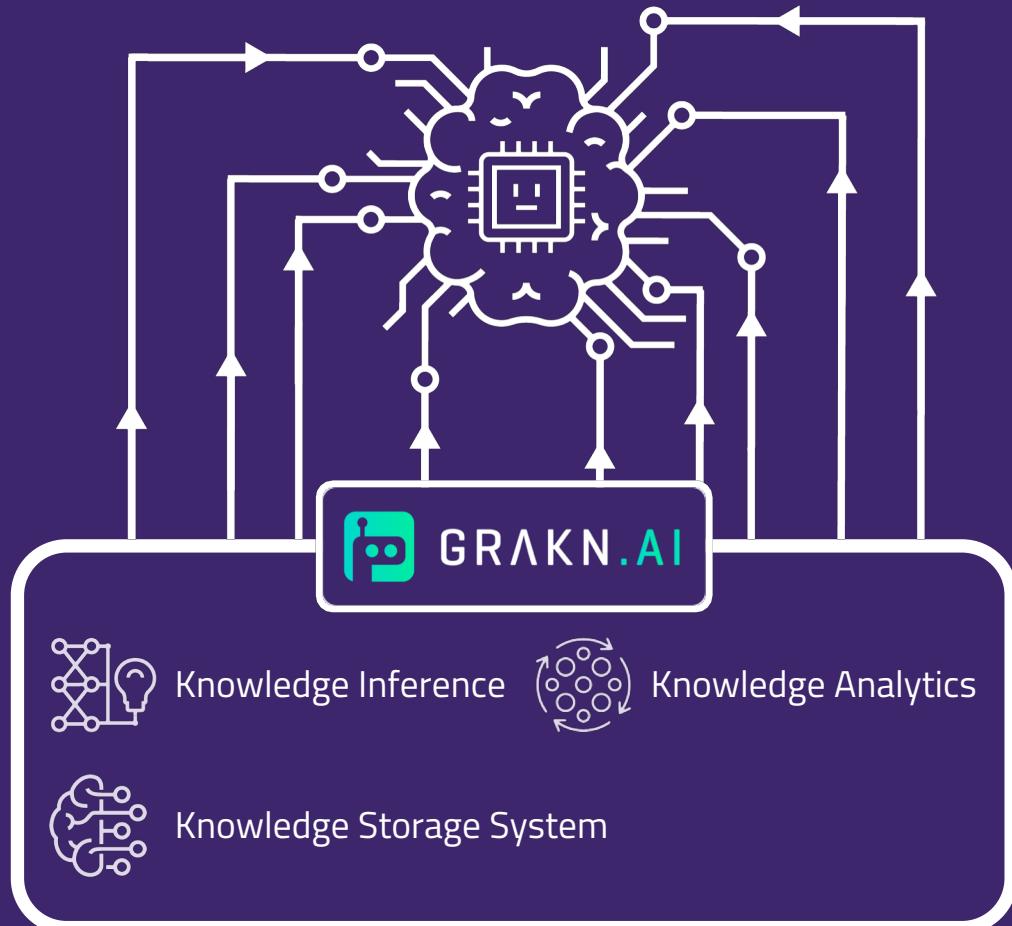
Use automated deductive reasoning to discover and interpret early diagnoses and recommend relevant therapeutic regimes



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# What Is Grakn?



GRAKN.AI is the knowledge base  
foundation for intelligent systems

a.k.a.

*"A KNOWLEDGE GRAPH"*



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DEMO...



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# How Do We Build A Precision Medicine Knowledge Graph?

1. Ingesting, integrating and normalising complex heterogeneous data.
2. Discovering and interpreting new insights.

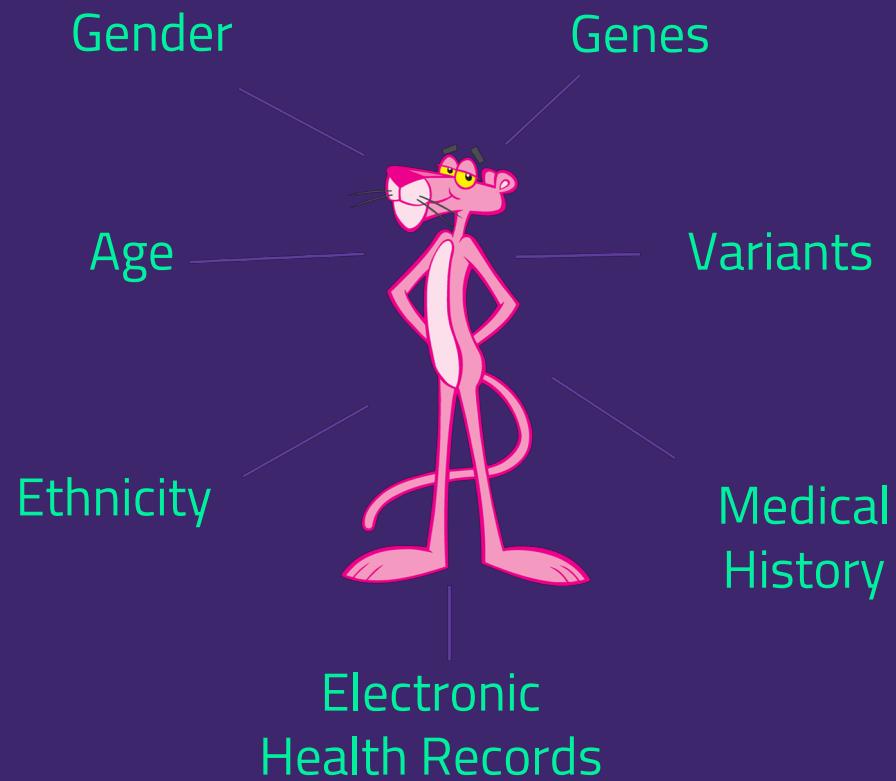


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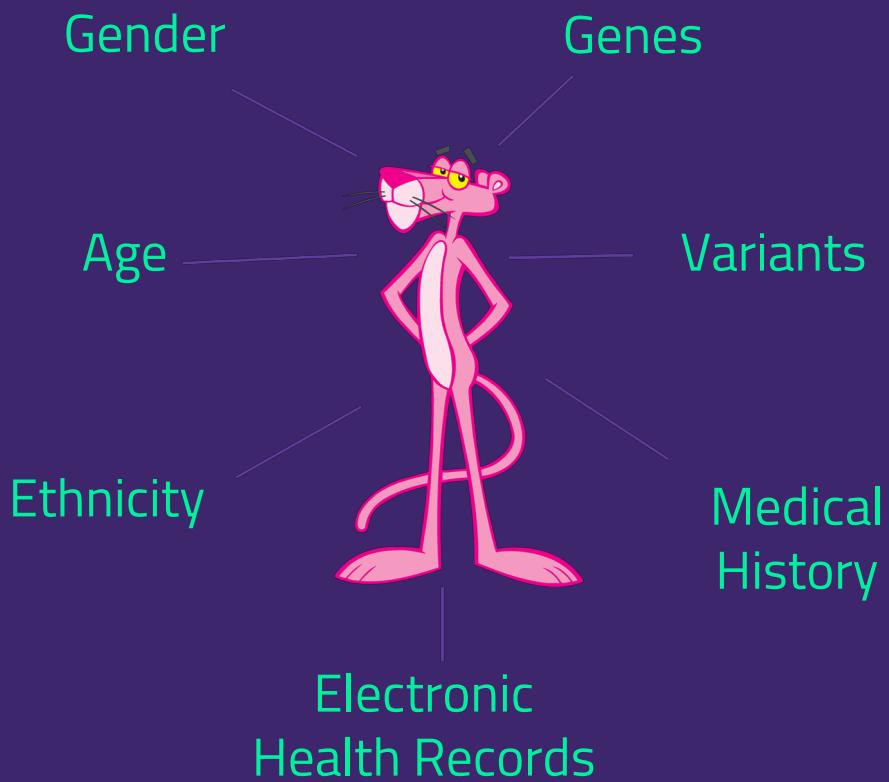


# Identify Data

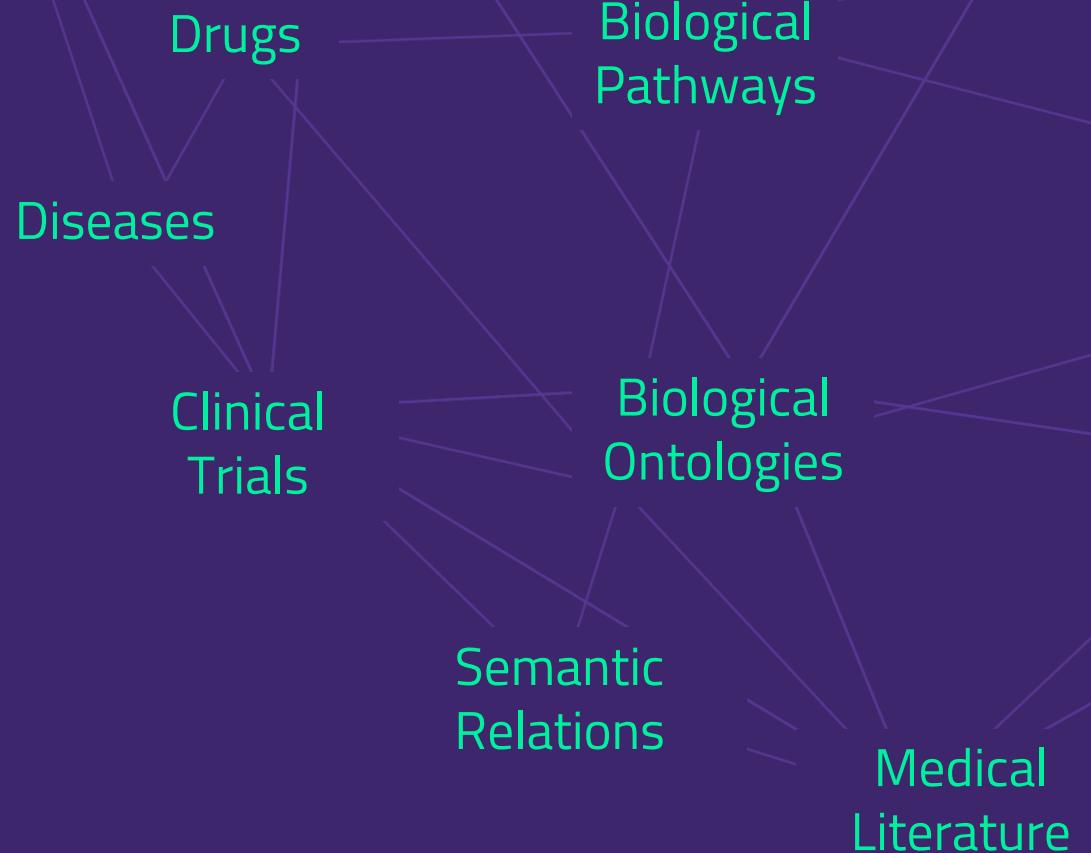


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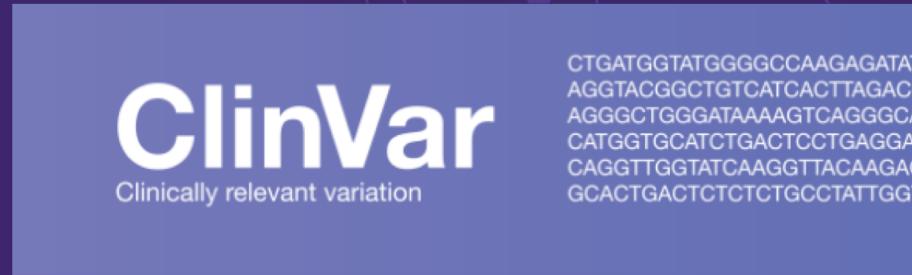
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## Identify Data



# Identify Data Sources



## OncOKB

## DRUGBANK

NIH U.S. National Library of Medicine  
*ClinicalTrials.gov*

# PubMed

# How Do We Normalise Raw Data?

People

person-id	age	gender	disease
...	...	...	...

Diseases

disease-id	name	type	...
...	...	...	...



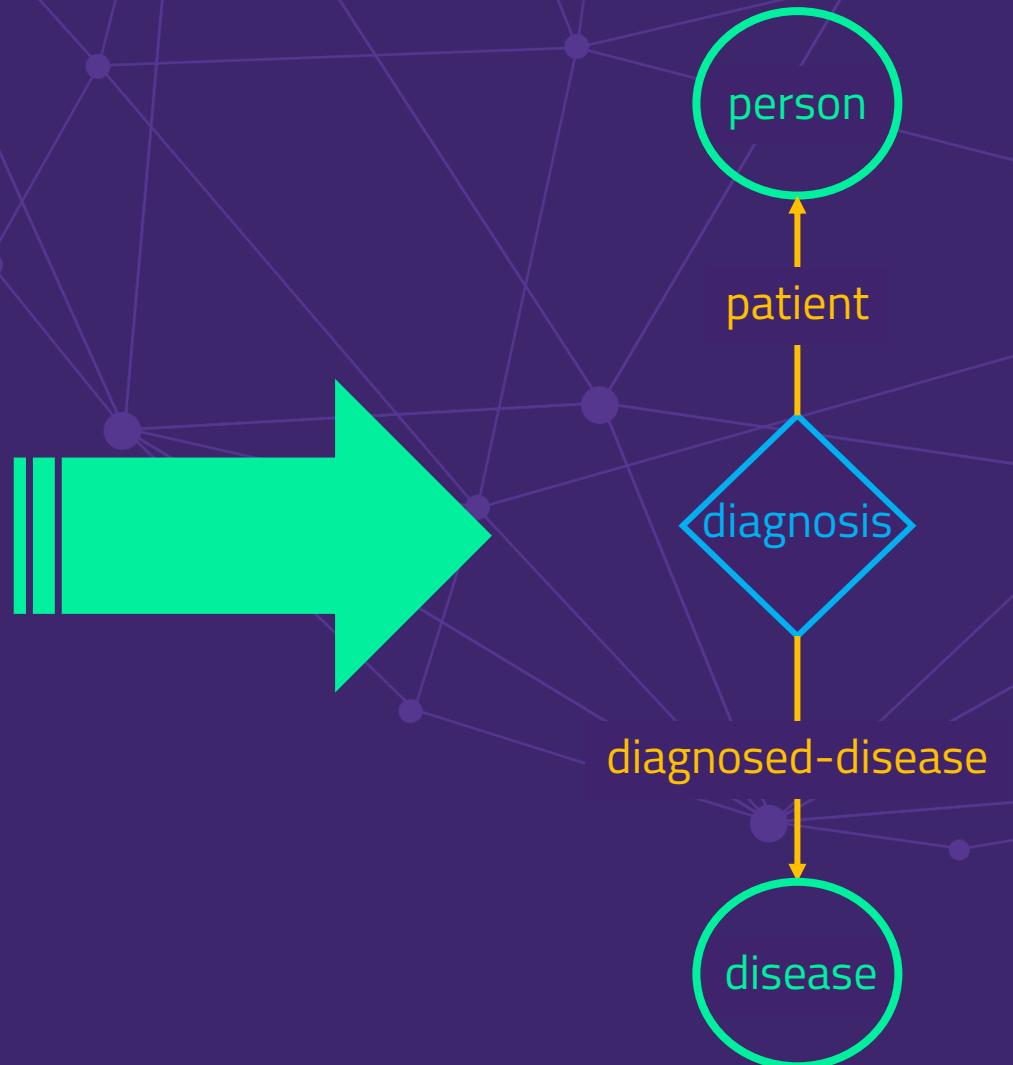
# How Do We Normalise Raw Data?

People

person-id	age	gender	disease
...	...	...	...

Diseases

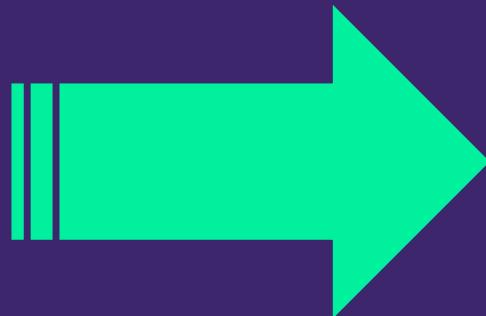
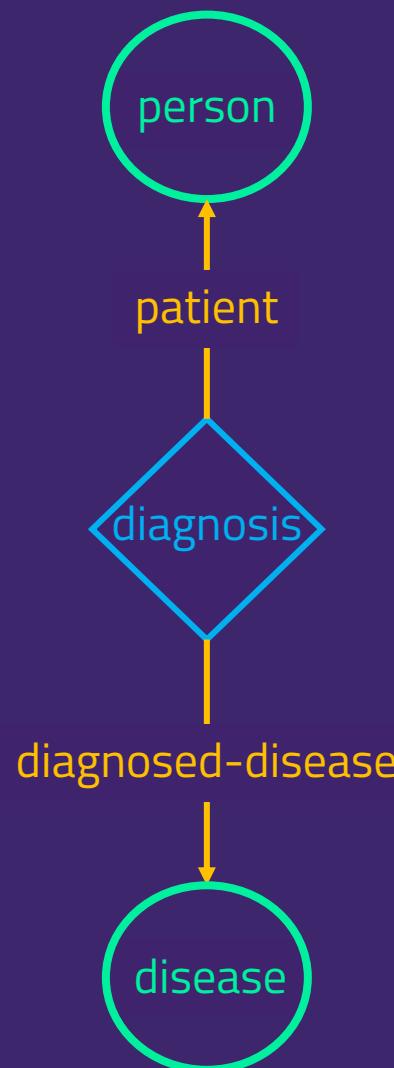
disease-id	name	type	...
...	...	...	...



# How Do We Represent The Normalisation In Graql?

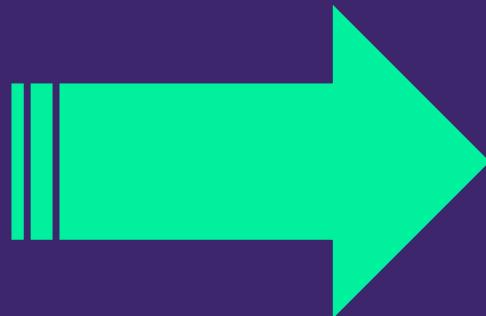
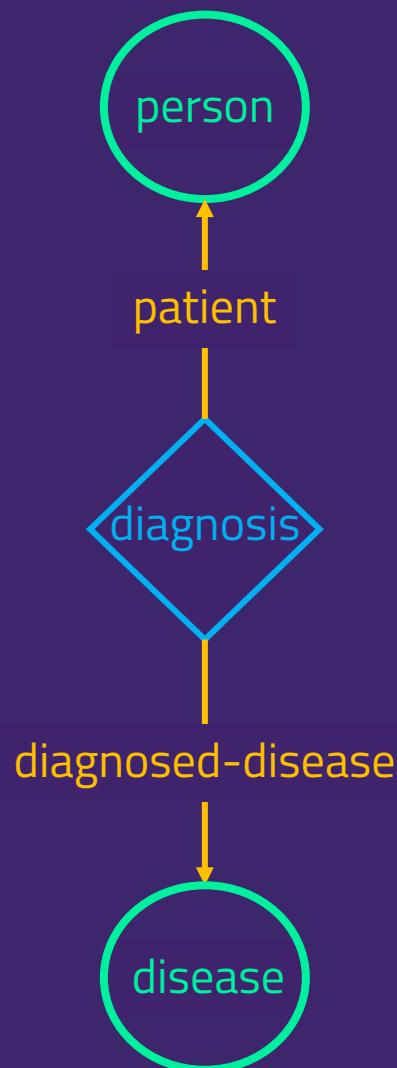


# How Do We Represent The Normalisation In Graql?



```
1 define
2
3 person sub entity,
4     has age,
5     has gender,
6     plays patient;
```

# How Do We Represent The Normalisation In Graql?



The window displays the following Graql code:

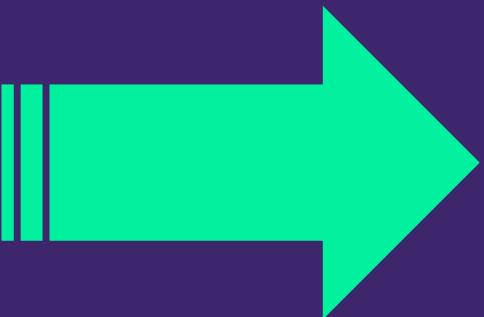
```
1 define
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3 person sub entity,
4   has age,
5   has gender,
6   plays patient;
7
8 disease sub entity,
9   has name,
10  has type,
11  plays diagnosed-disease;
```

# How Do We Represent The Normalisation In Graql?



```
1 define
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3 person sub entity,
4   has age,
5   has gender,
6   plays patient;
7
8 disease sub entity,
9   has name,
10  has type,
11  plays diagnosed-disease;
12
13 diagnosis sub relationship,
14   relates patient,
15   relates diagnosed-disease;
```

# How Do We Represent The Normalisation In Graql?



```
1 define
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3 person sub entity,
4   has age,
5   has gender,
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8 disease sub entity,
9   has name,
10  has type,
11  plays diagnosed-disease;
12
13 diagnosis sub relationship,
14   relates patient,
15   relates diagnosed-disease;
16
17 age sub attribute datatype double;
18 gender sub attribute datatype string;
19 name sub attribute datatype string;
20 type sub attribute datatype string;
```

# How Do We Migrate Data Into Grakn?

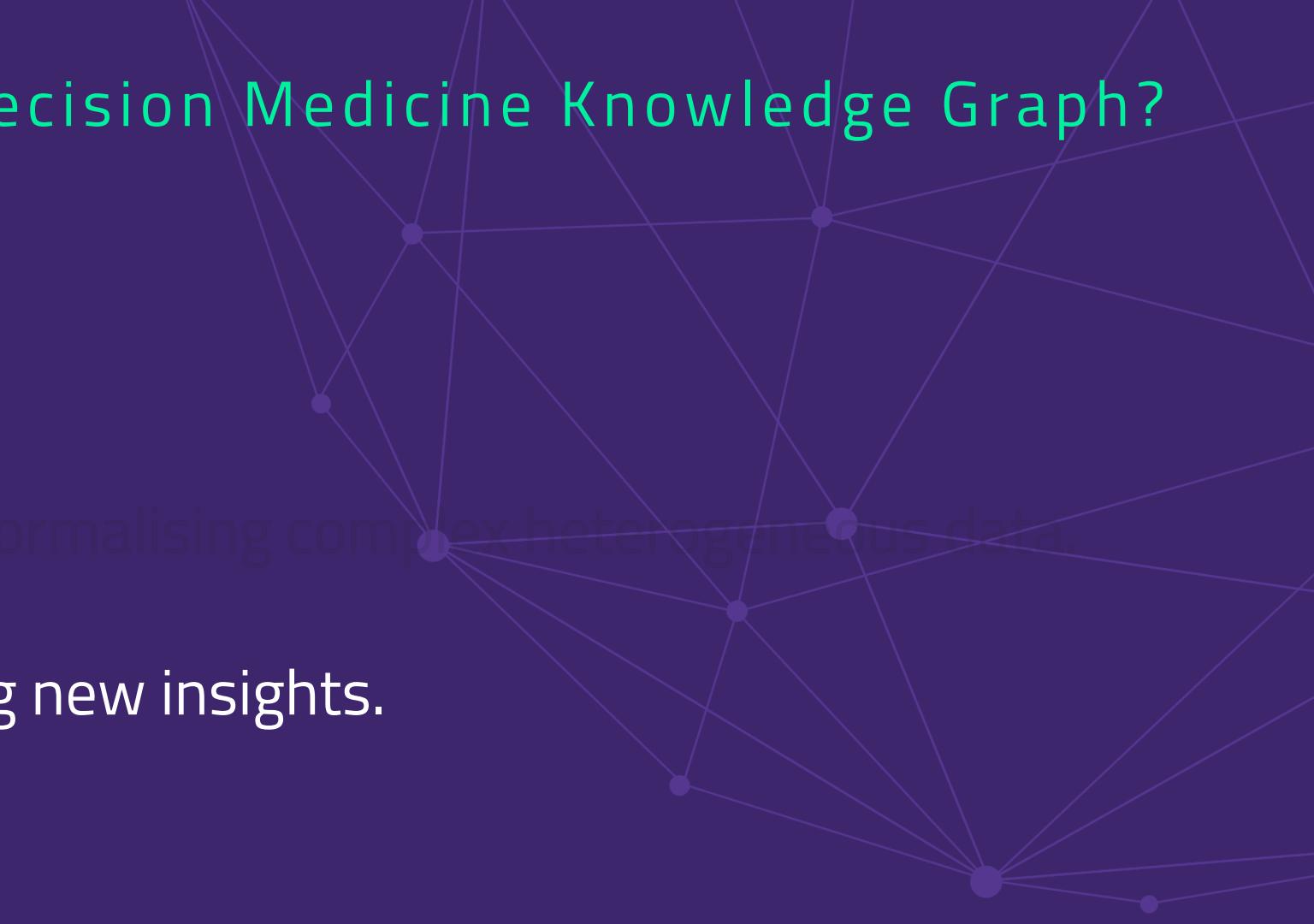


```
InsertQuery query = Graql.insert(  
    var("d").isa("disease").has("name", "melanoma").has("type", "cancer")  
);
```

```
const query = await transaction.query (  
    "insert $d isa disease, has name \"melanoma\", has type \"cancer\";"  
);
```

```
query = transaction.query(  
    "insert $d isa disease, has name \"melanoma\", has type \"cancer\";"  
)
```

# How Do We Build A Precision Medicine Knowledge Graph?

- 
1. Ingesting, integrating and normalising complex heterogeneous data.
  2. Discovering and interpreting new insights.



# How Do We Discover An Insight?

Question

Given a person; what are the diseases she/he can be potentially diagnosed with?



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# How Do We Discover An Insight?

Question

Given a **person**; what are the **diseases** she/he can be potentially diagnosed with?

=

Graql

```
1 match $p isa person;  
2 $d isa disease;  
3 $r ($p, $d) isa early-diagnosis;  
4 get; limit 1;
```



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# How Do We Discover An Insight?

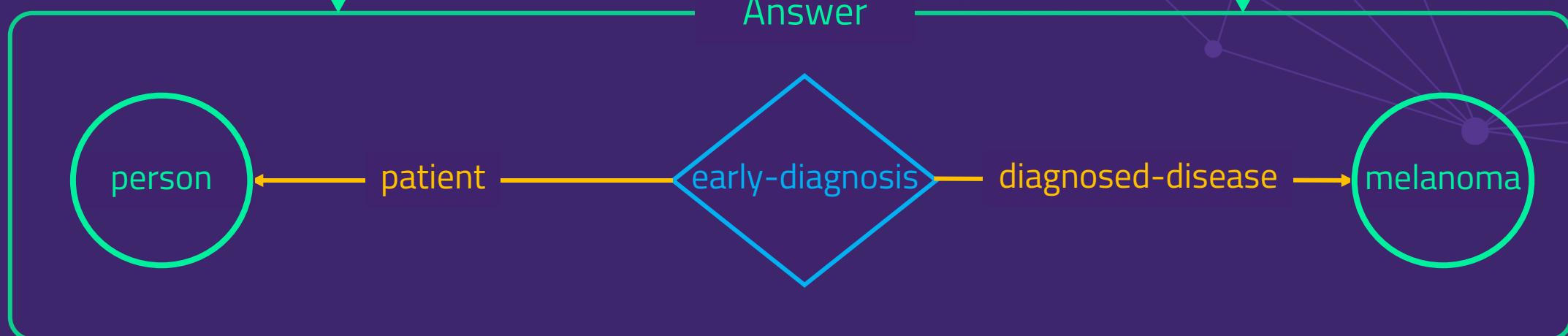
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# How Do We Discover An Insight?

## Question

Given a person suffering from melanoma; which clinical trials could be recommended to her/him?



# How Do We Discover An Insight?

Question

Given a **person** suffering from **melanoma**; which **clinical trials** could be recommended to her/him?

=

Graql

```
1 match $p isa person;
2 $d isa disease has disease-name "melanoma";
3 ($p, $d) isa diagnosis;
4 $c isa clinical-trial;
5 $r ($p, $c) isa personalised-therapy;
6 get; limit 1;
```



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# How Do We Discover An Insight?

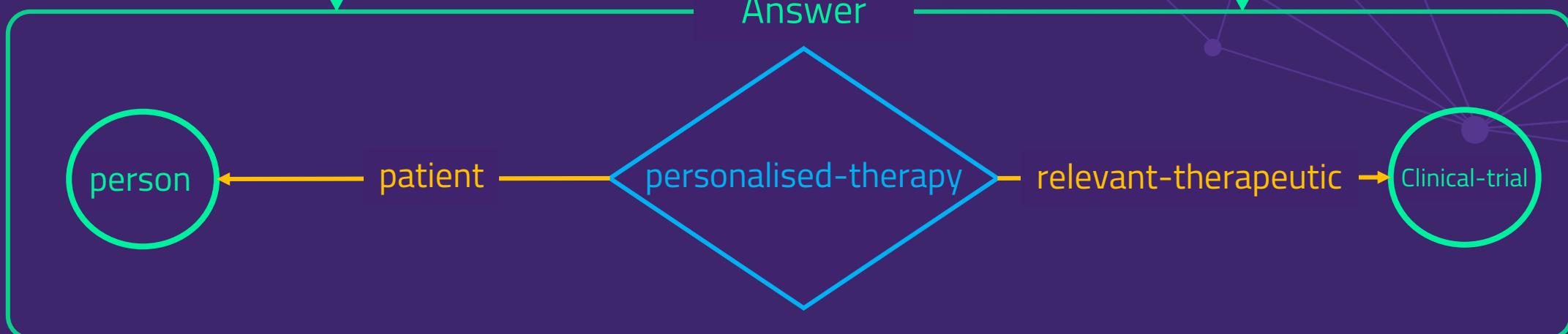
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Given a **person** suffering from **melanoma**; which **clinical trials** could be recommended to her/him?

=

Graql

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# How Do We Interpret Insights?

Explanation

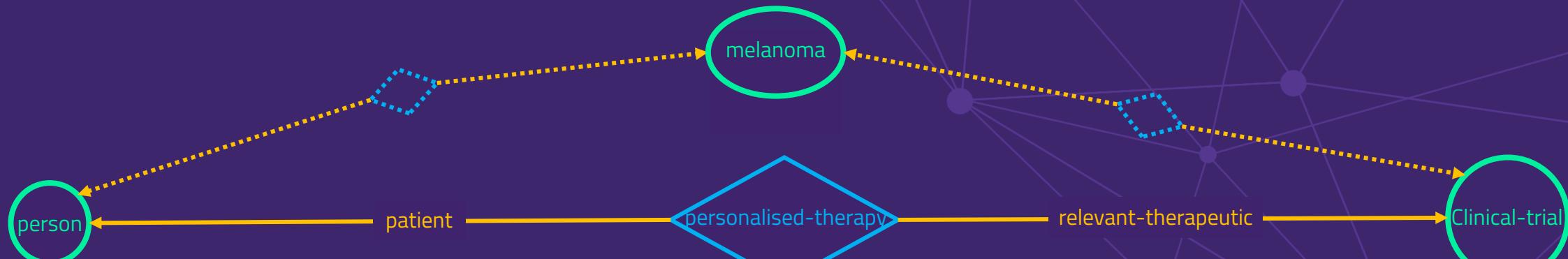


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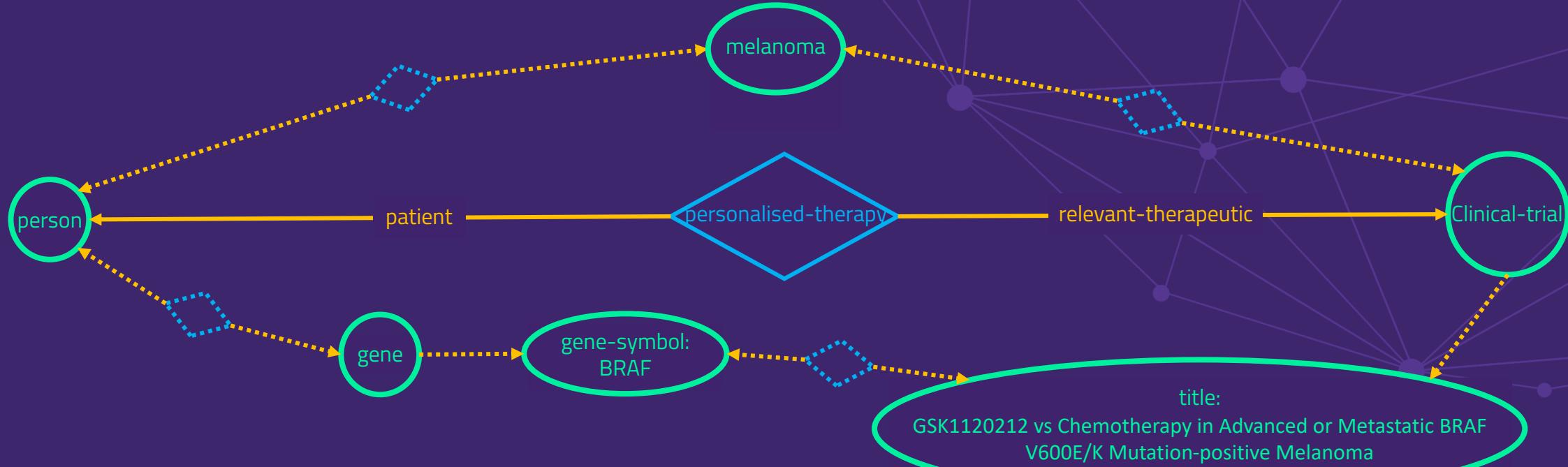
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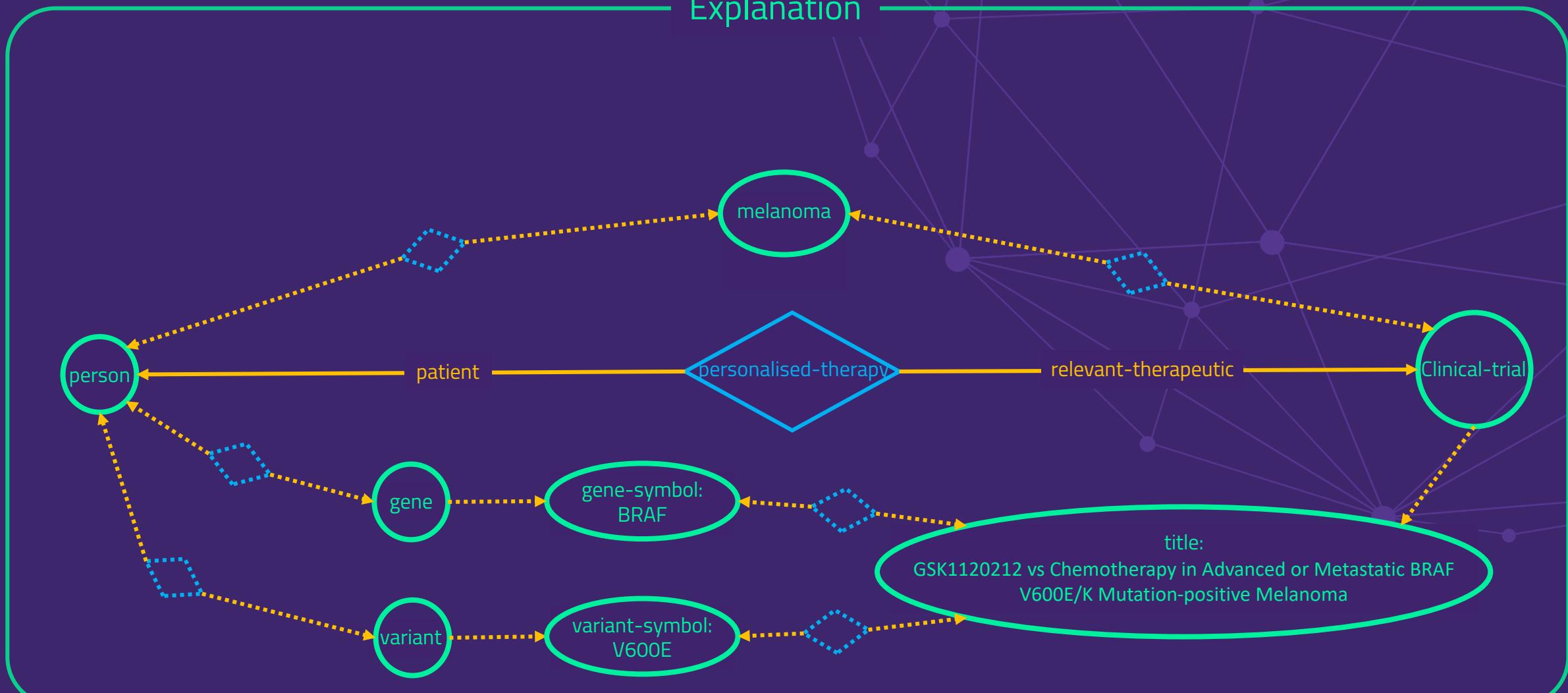


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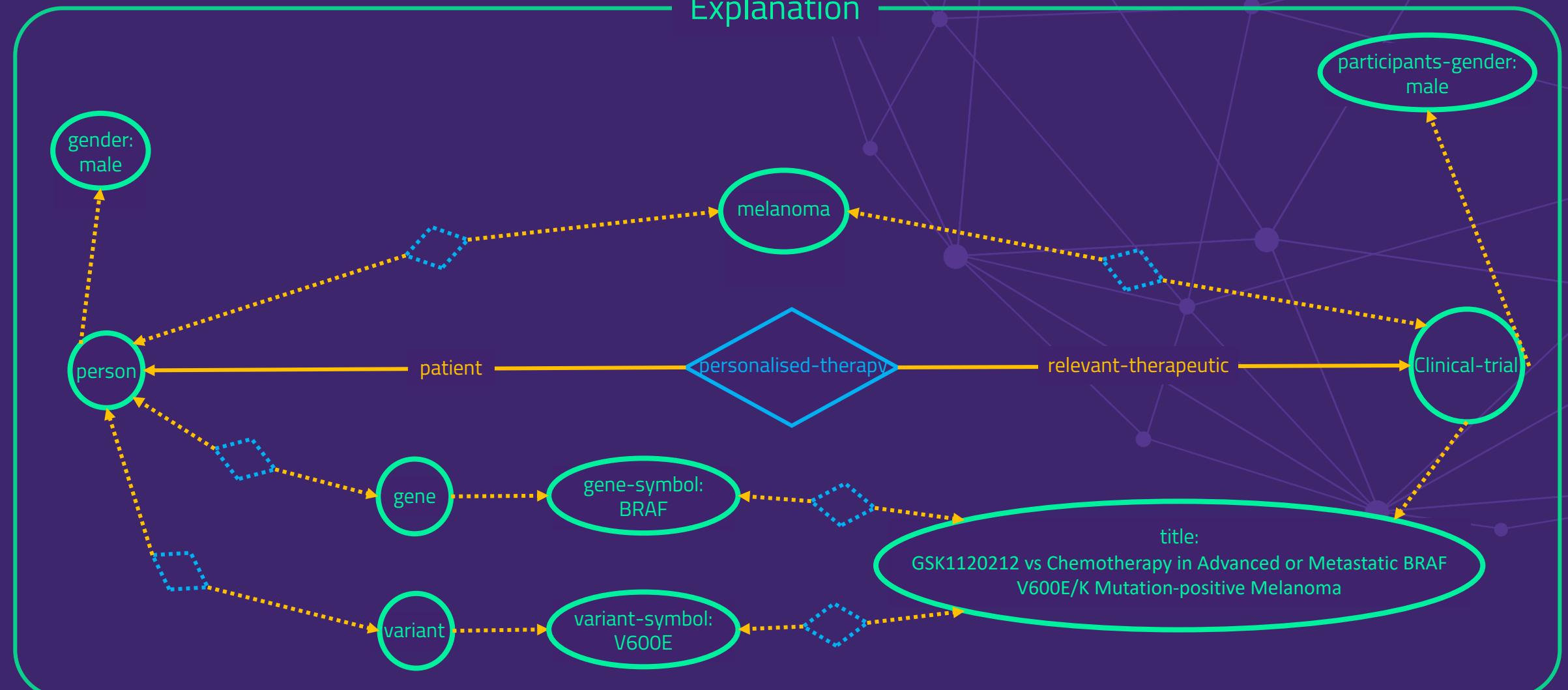
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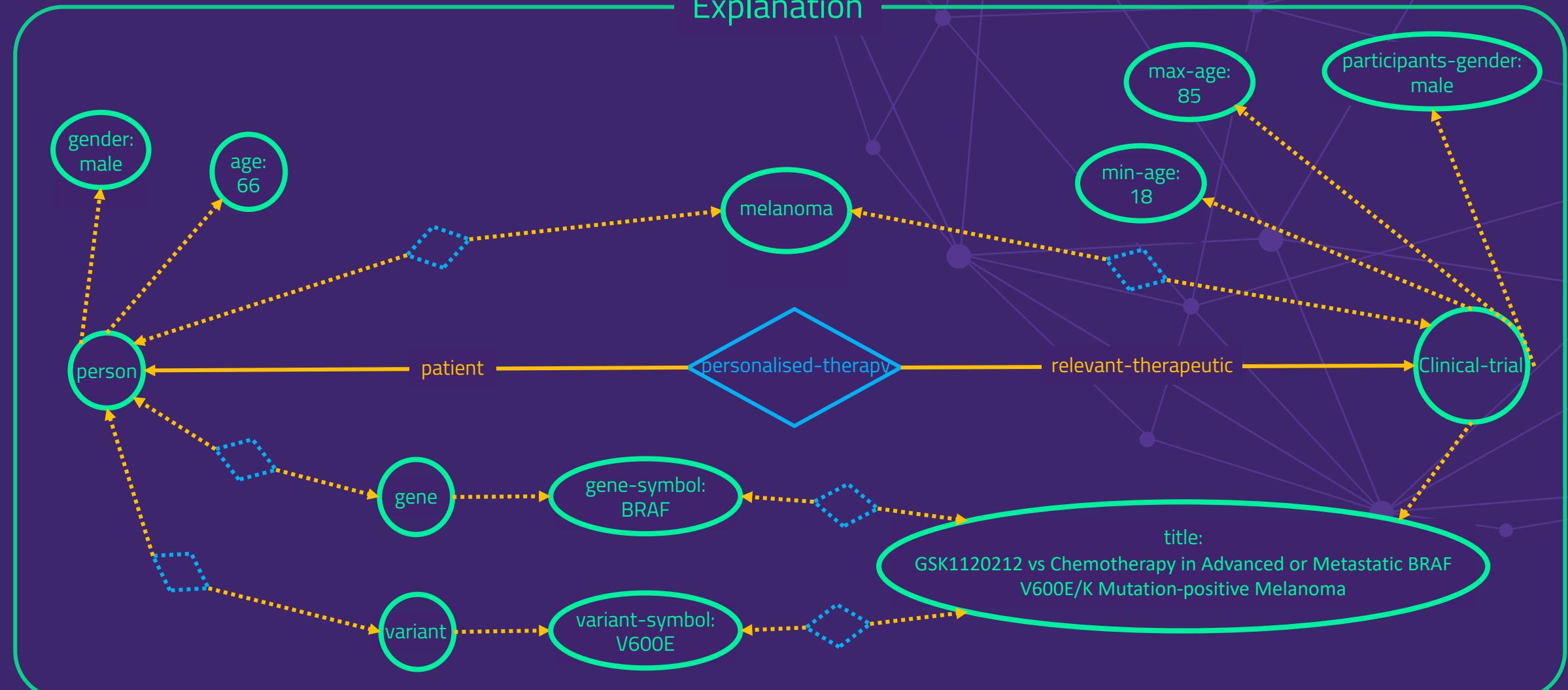


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# How Do We Interpret Insights?

Explanation



# How Do We Represent Reasoning in Graql?

```
1 define
2
3 personalised-patient-therapy sub rule
4 when {
5     ($person, $trial) isa eligible-trial-participant;
6     ($person, $trial) isa relevant-trial-participant;
7 }
8 then {
9     ($person, $trial) isa personalised-therapy;
10 };
```



# How Do We Represent Reasoning in Graql?

```
11 trial-participant-eligibility sub rule,
12 when {
13     $person isa person, has age $age, has gender $gender;
14     $trial isa clinical-trial,
15     has min-age <= $age,
16     has max-age >= $age,
17     has gender == $gender;
18     $disease isa disease; ($disease, $person); ($disease, $trial);
19 },
20 then {
21     ($person, $trial) isa eligible-trial-participant;
22 };
```

```
1 define
2
3 personalised-patient-therapy sub rule
4 when {
5     ($person, $trial) isa eligible-trial-participant;
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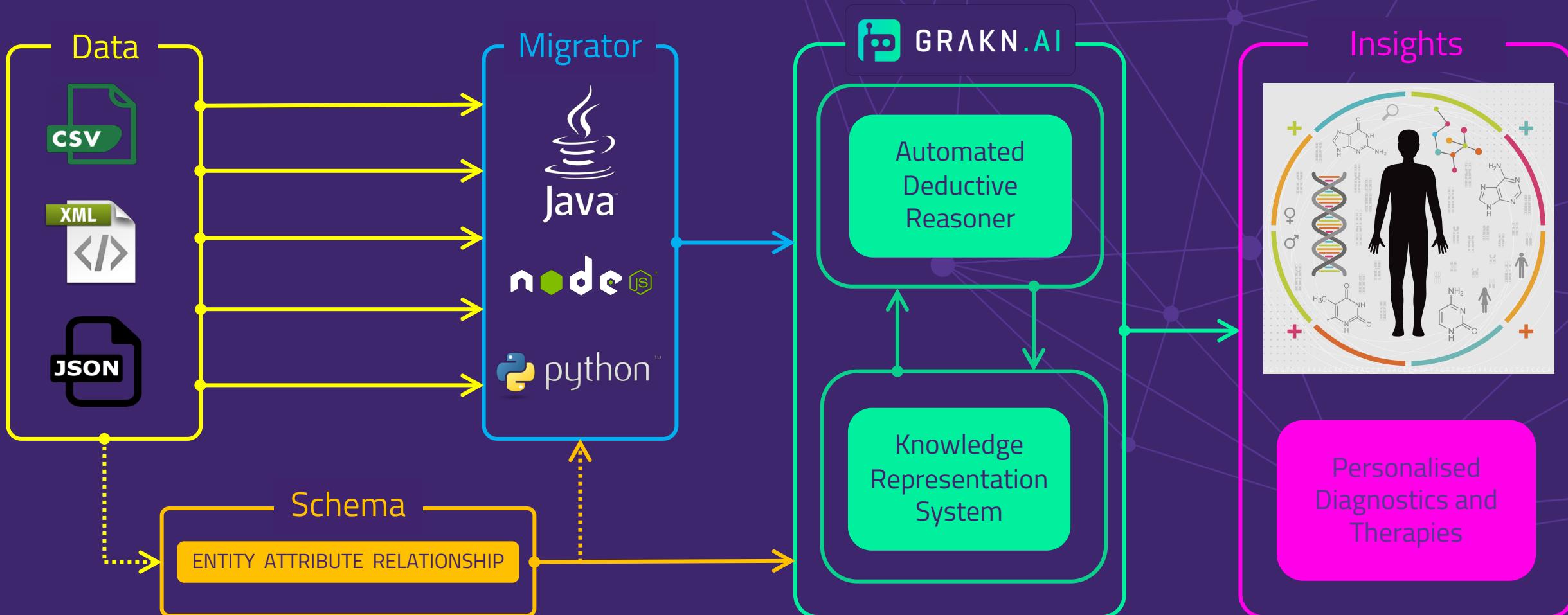
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11 trial-participant-eligibility sub rule,
12 when {
13     $person isa person, has age $age, has gender $gender;
14     $trial isa clinical-trial,
15     has min-age <= $age,
16     has max-age >= $age,
17     has gender == $gender;
18     $disease isa disease; ($disease, $person); ($disease, $trial);
19 },
20 then {
21     ($person, $trial) isa eligible-trial-participant;
22 };
```

```
1 define
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3 personalised-patient-therapy sub rule
4 when {
5     ($person, $trial) isa eligible-trial-participant;
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7 }
8 then {
9     ($person, $trial) isa personalised-therapy;
10 };
```

```
23 trial-participant-relevance sub rule,
24 when {
25     $person isa person,
26     ($person, $gene); $gene isa gene, has symbol $gs;
27     ($person, $variant); $variant isa variant, has symbol $vs;
28     $trial isa clinical-trial, has title contains $gs, has title contains $vs;
29 },
30 then {
31     ($person, $trial) isa relevant-trial-participant;
32 };
```



# Architecture



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# Precision Medicine Knowledge Graph



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## Normalisation

Impose an explicit structure on the data to contextualise the relationships within multi-omics and patient networks

## Discovery

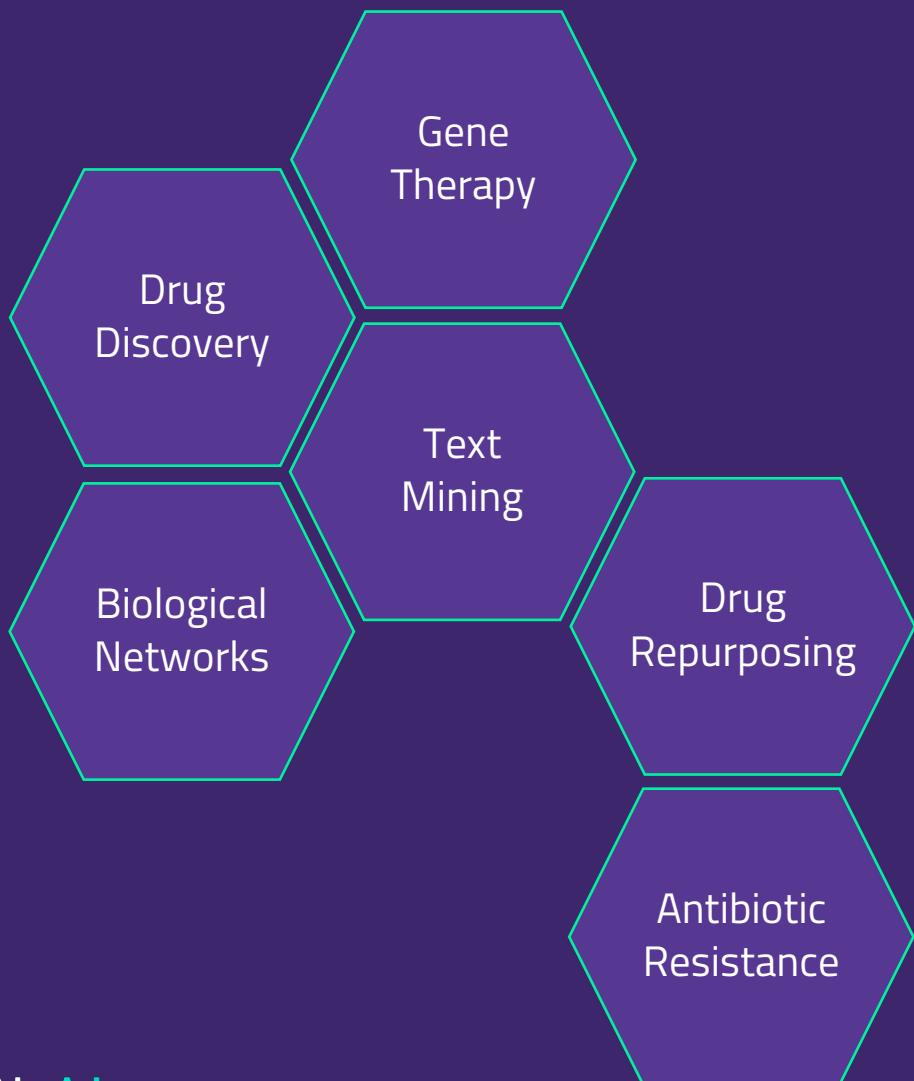
Use automated deductive reasoning to discover and interpret early diagnoses and recommend relevant therapeutic regimes



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# What Else Can We Use Grakn For In Bio-Medicine?





Questions?

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