National Institute of Environmental Health Sciences
Your Environment. Your Health.

"Making Connections that Matter: Harnessing Knowledge Graphs for Biomedical Discovery and Integration"

Darius Bost, PhD

Health Data Scientist [C]

Too Much Data, Not Enough Connection

50 YEARS OMIM
Human Genetics Knowledge for the World

```
>NG_008679.1:5001-38170 Homo sapiens paired box 6 (PAX6)
ACCCTCTTTTCTTATCATTGACATTTAAACTCTGGGGCAGGTCCTCGCGTAGAACGCGGCTGTCAGATCT
GCCACTTCCCCTGCCGAGCGGCGGTGAGAAGTGTGGGAACCGGCGCTGCCAGGCTCACCTGCCTCCCCGC
CCTCCGCTCCCAGGTAACCGCCCGGGCTCCGGCCCCGGCCCGGCTCGGGGCCCGCGGGGCCTCTCCGCTG
CCAGCGACTGCTGTCCCCAAATCAAAGCCCGCCCCAAGTGGCCCCGGGGCTTGATTTTTGCTTTTAAAAG
GAGGCATACAAAGATGGAAGCGAGTTACTGAGGGAGGGATAGGAAGGGGGGTGGAGGAGGGACTTGTCTT
TGCCGAGTGTGCTCTTCTGCAAAAGTAGCAAAATGTTCCACTCCTAAGAGTGGACTTCCAGTCCGGCCCT
GAGCTGGGAGTAGGGGGCGGGAGTCTGCTGCTGCTGTCTGCTAAAGCCACTCGCGACCGCGAAAAATGCA
GGAGGTGGGGACGCACTTTGCATCCAGACCTCCTCTGCATCGCAGTTCACGACATCCACGCTTGGGAAAG
TCCGTACCCGCGCCTGGAGCGCTTAAAGACACCCTGCCGCGGGTCGGGCGAGGTGCAGCAGAAGTTTCCC
GCGGTTGCAAAGTGCAGATGGCTGGACCGCAACAAAGTCTAGAGATGGGGTTCGTTTCTCAGAAAGACGC
```

FIGURE 7.1: An example fasta file showing the first part of the PAX6 gene.

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1799 2025

1 Cite Share

Understanding the immunology of **asthma**: Pathophysiology, biomarkers, and treatments for **asthma** endotypes.

Gans MD, Gavrilova T.

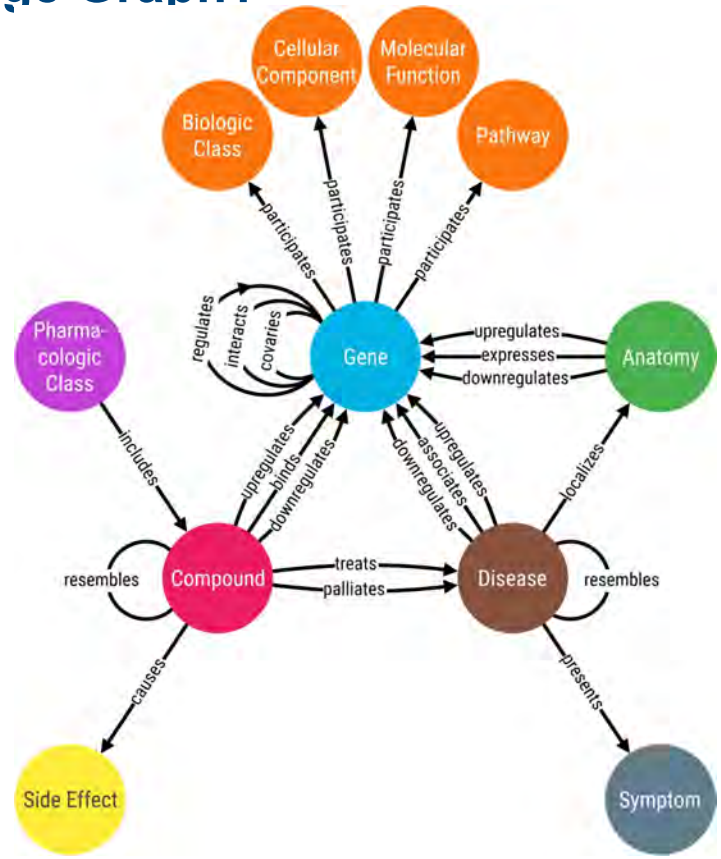
Paediatr Respir Rev. 2020 Nov;36:118-127. doi: 10.1016/j.prrv.2019.08.002. Epub 2019 Oct 9.

PMID: 31678040 Review.

Asthma is now recognized as a heterogeneous disease and emerging clinical and laboratory research has elucidated understanding of **asthma**'s underlying immunology. ...By understanding the immunology of **asthma**, practitioners will be able to diagnose patie ...

"How do I link gene expression data with clinical outcomes and drug targets?"

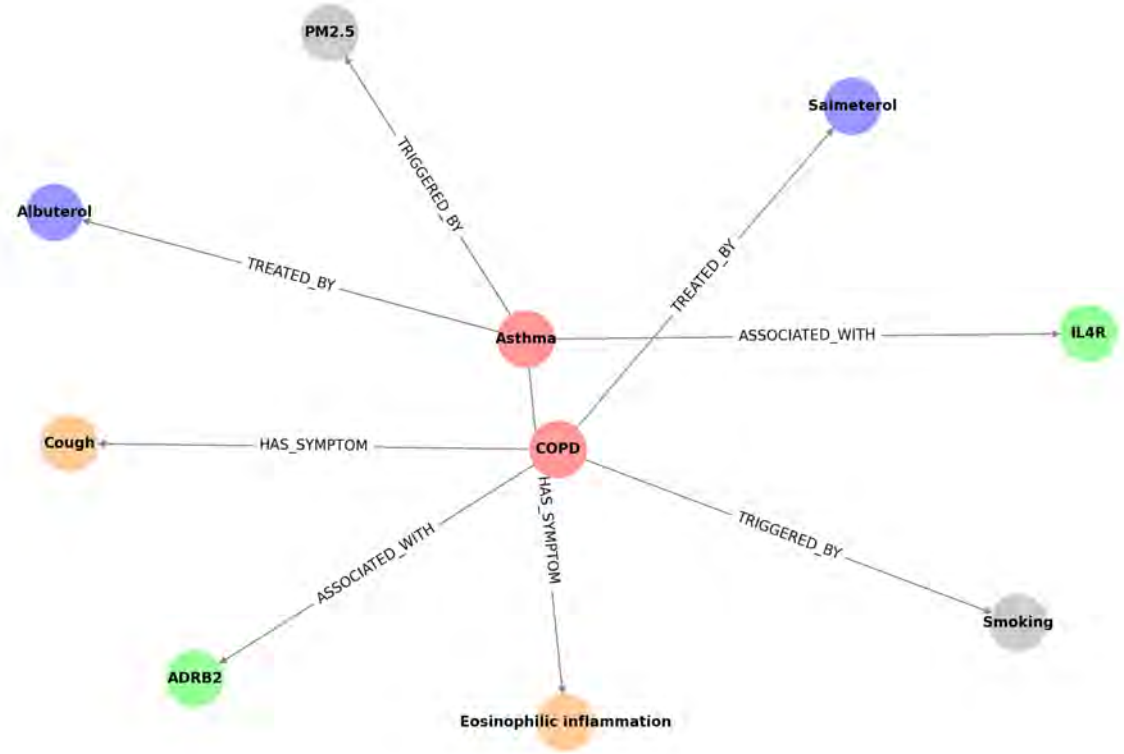
What Is a Knowledge Graph?



How KGs Differ from Tables

Disease	Gene	Drug	Environment	Symptom
Asthma	IL4R	Albuterol	PM2.5	Eosinophilic inflammation
COPD	ADRB2	Salmeterol	Smoking	Cough

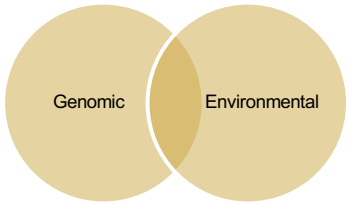
How KGs Differ from Tables



What Can a KG Do for You?

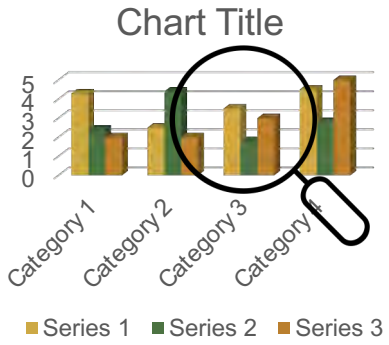
Cross-Domain Integration

Unify clinical, genomic, environmental, and chemical data.



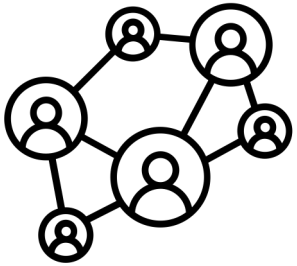
Flexible Querying

Ask complex, multi-hop questions.



Expandable & FAIR

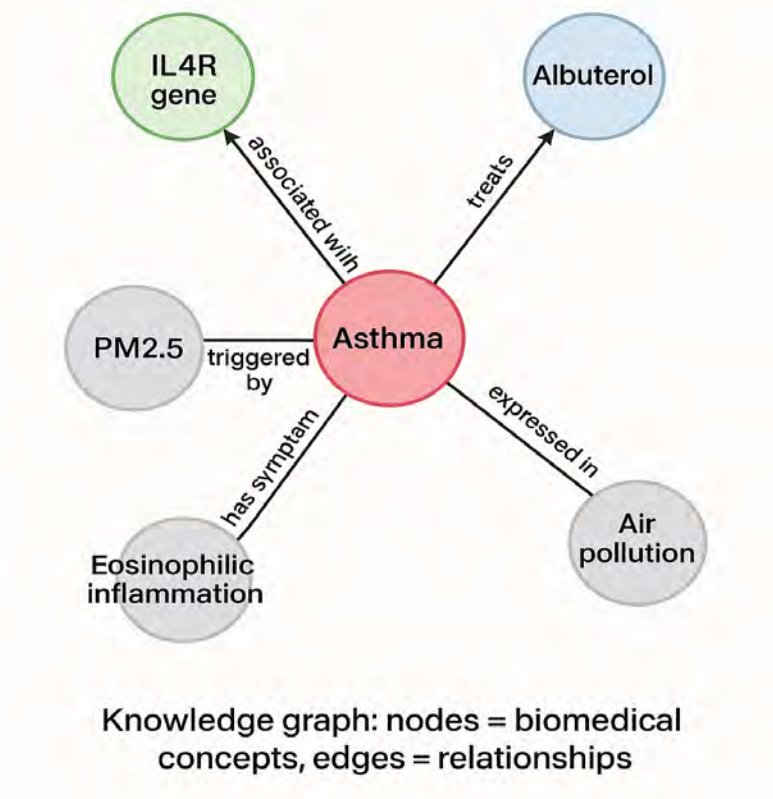
Supports updates, ontologies, and standards.



Hypothesis Generation

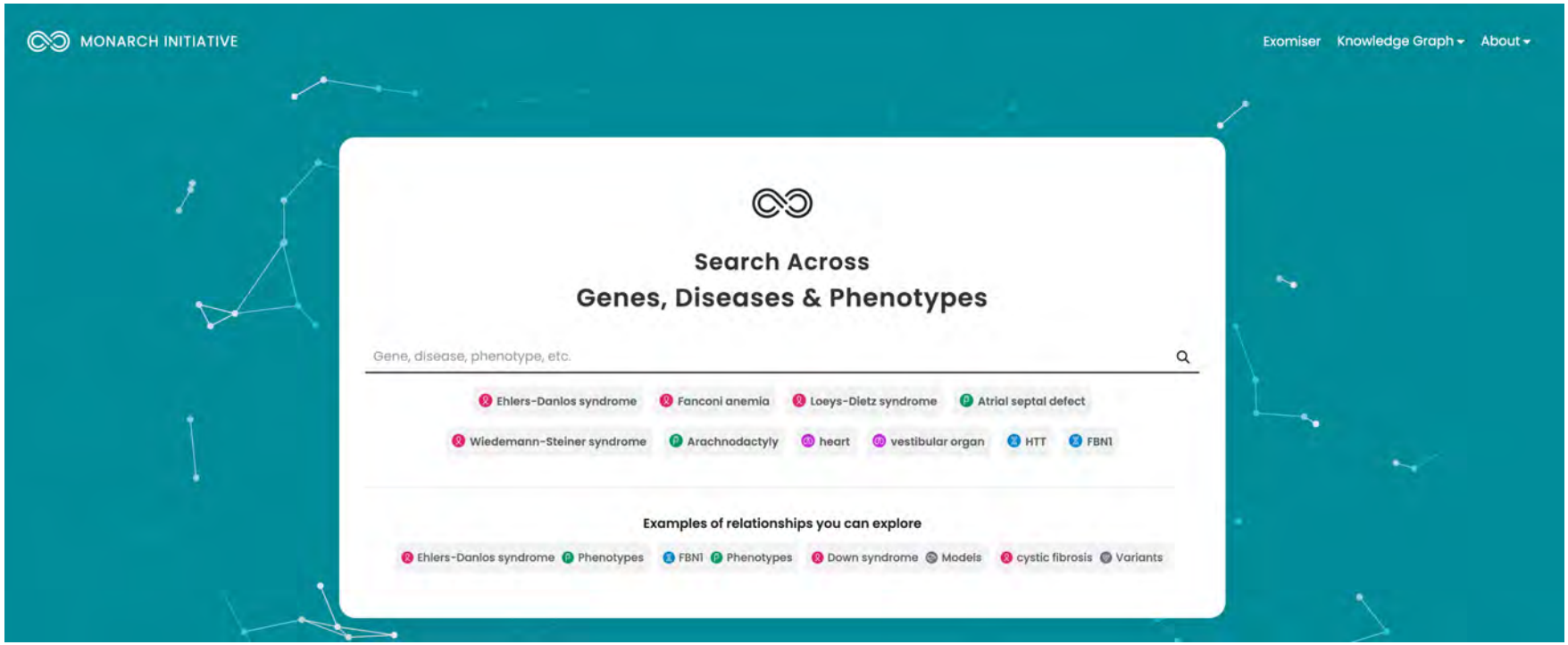
Reveal non-obvious connections in your data.

Use Case Snapshot:
Example: Linking Asthma, Genes, and Environment



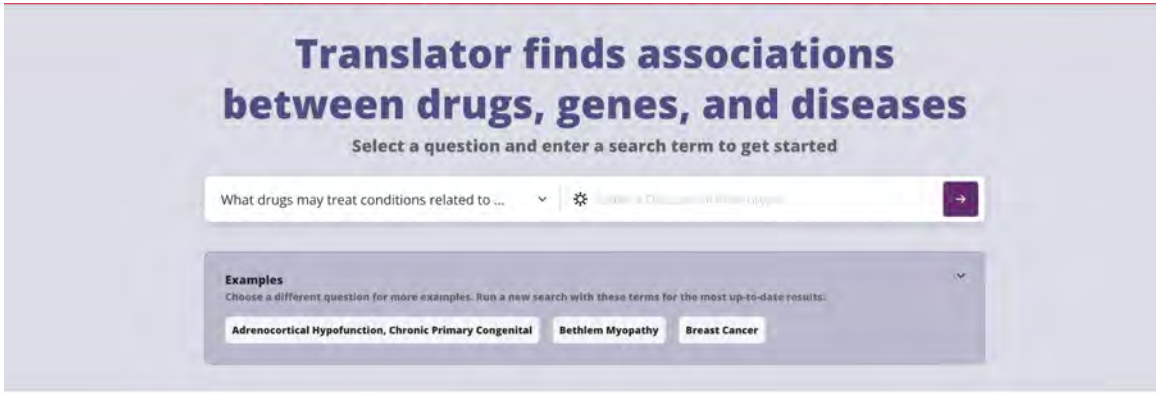
Knowledge graph: nodes = biomedical concepts, edges = relationships

Demo 1 – Monarch Initiative



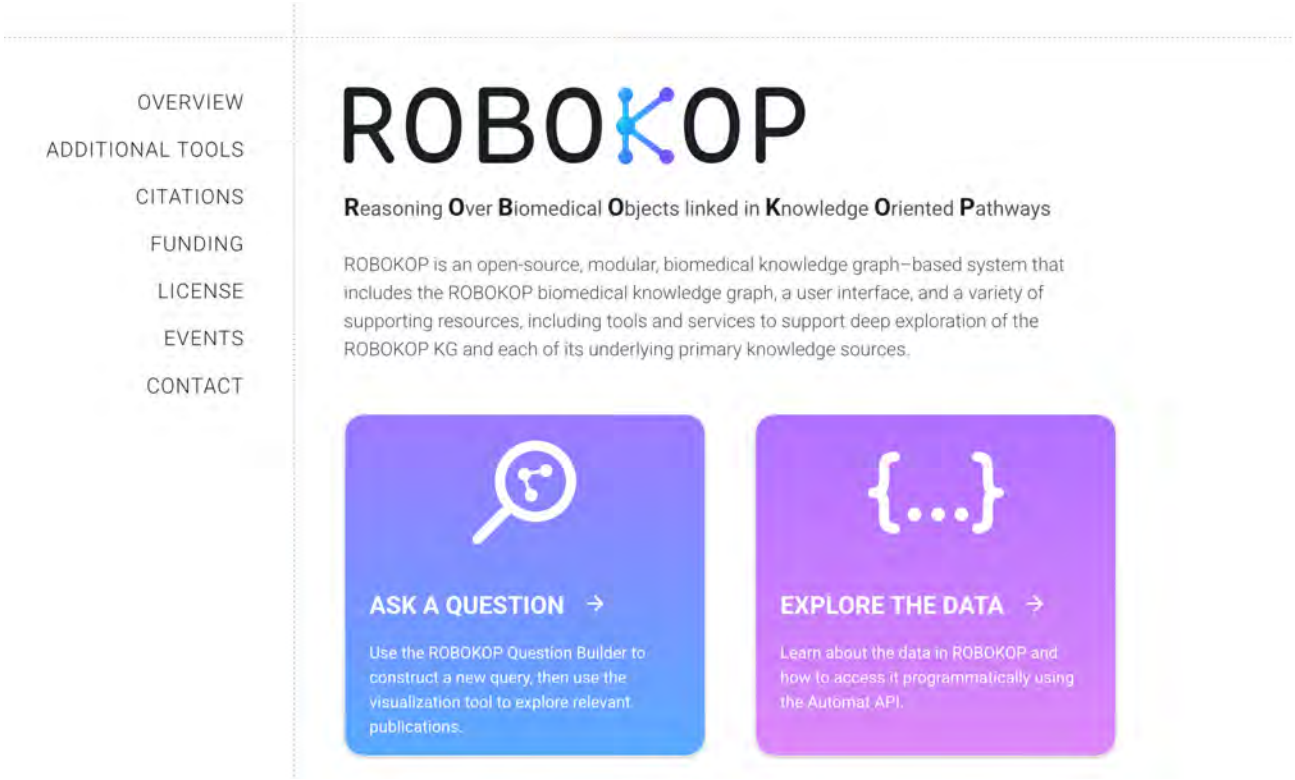
https://monarchinitiative.org/

Demo 2 –NCATS Biomedical Translator



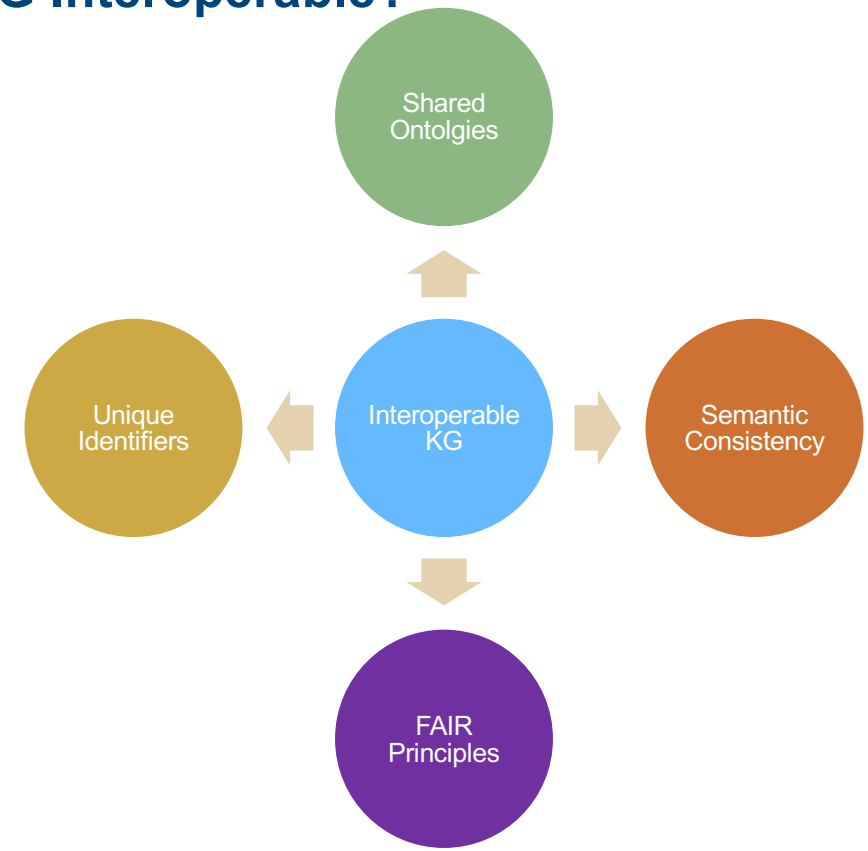
https://ui.transltr.io/

Demo 3- ROBOKOP: Asking Biomedical Questions Through Graphs



https://robokop.renci.org/

What Makes a KG Interoperable?



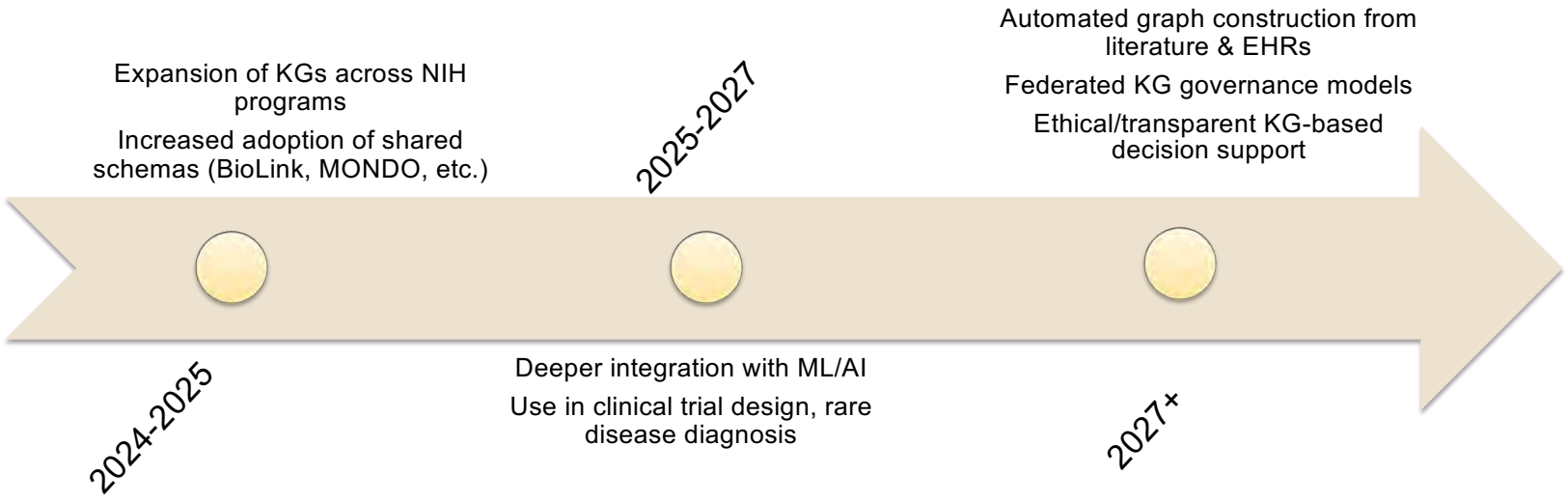
Why Interoperability Matters

"When we make knowledge graphs interoperable, we unlock major benefits:

- ✅ Enables **scalable integration**
- ✅ Enhances **reproducibility**
- ✅ Supports **collaboration**
- ✅ Promotes **data reuse**

🧩 **Scalable Integration**	Combine diverse datasets (e.g., EHRs + GWAS + exposures) seamlessly
🔁 **Reproducibility**	Standard terms make results testable and repeatable
🤝 **Collaboration**	Graphs can be shared across teams and institutions
🔄 **Data Reuse**	One KG, many questions — from research to clinical application

The Road Ahead for Biomedical Knowledge Graphs



From Data to Discovery

- Knowledge graphs connect biomedical domains into a computable network
- Tools are available for non-technical users (Monarch, Translator, ROBOKOP)
- Interoperability enables scale, reuse, and collaboration
- The future includes AI, clinical applications, and governance

Questions & Discussion

darius.bost@nih.gov