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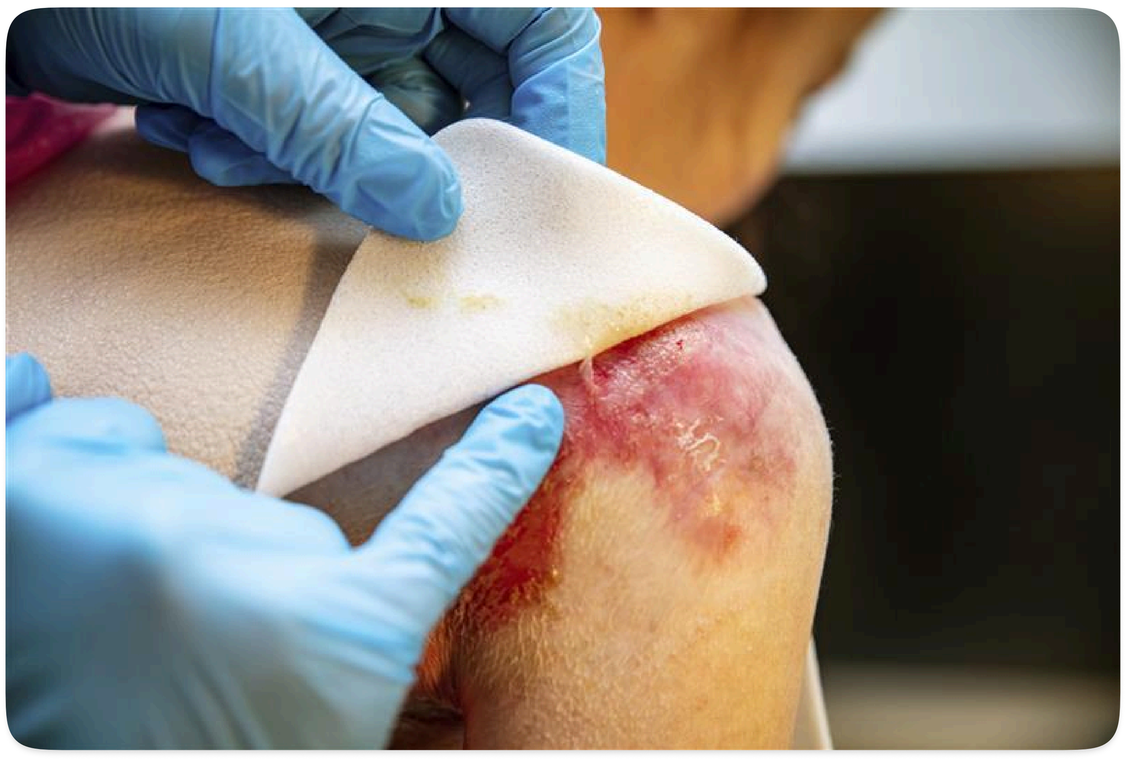
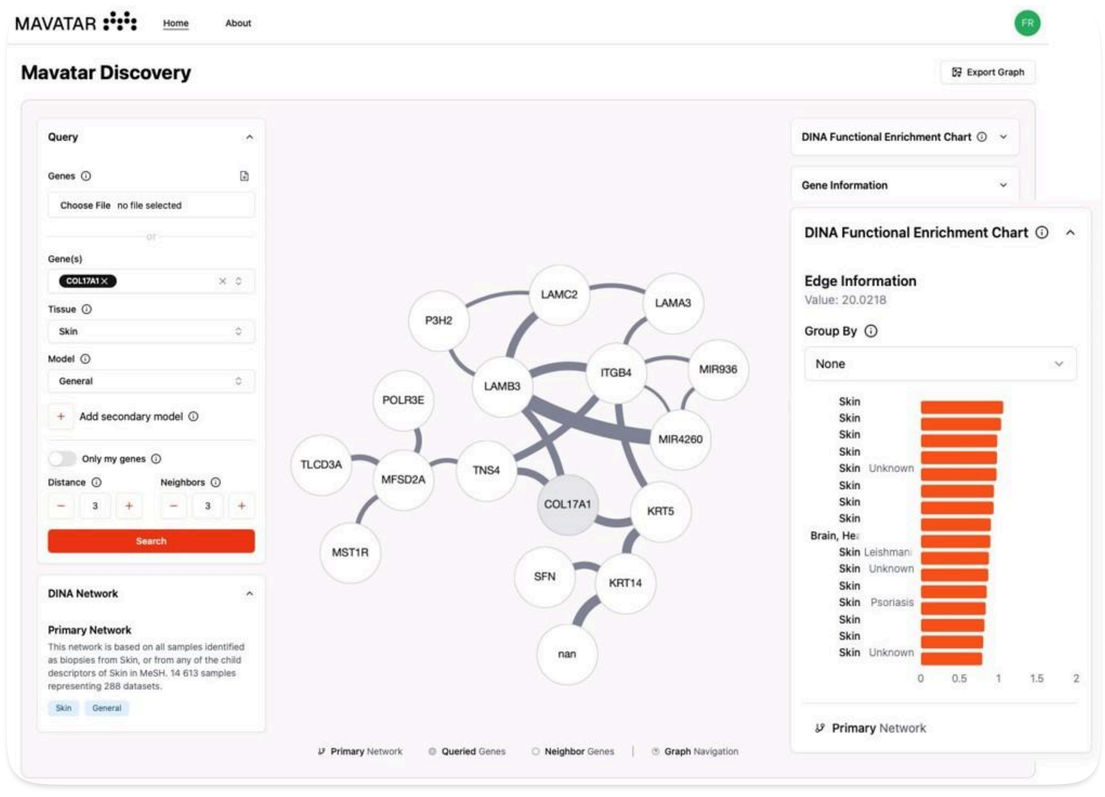
Case Study: Mapping Disease Pathways in Epidermolysis Bullosa Using Mavatar Discovery

Background

Epidermolysis Bullosa (EB) is a rare genetic disorder characterized by extreme skin fragility, chronic wounds, and high cancer risk. It is caused by mutations in genes encoding structural proteins like collagens and keratins. While 15 EB-associated genes are known, researchers face challenges linking them into functional networks due to the scarcity of disease-specific transcriptomic data.





Challenge

Even though the genetic basis of EB is partially known, it remains difficult to reconstruct a complete molecular disease pathway. Conventional tools lack the integration of tissue-specific context and fail to reveal functional connections among known and emerging genes, limiting the identification of therapeutic targets or modifier pathways.

Solution

Using Mavatar Discovery, researchers analyzed EB genes like COL17A1 within a skin-specific transcriptomic framework. By leveraging integrated data from related skin conditions (e.g., wounds, psoriasis, dermatitis), they built co-expression networks centered on COL17A1. The platform's local enrichment analysis identified clusters of EB genes and their biological neighbors, many of which were previously unlinked to EB.

Results

The analysis revealed two core disrupted systems in EB: laminin/integrin complexes and keratin structures. Notably, 6 of the 17 known EB genes were significantly enriched in a single gene neighborhood — a >400x enrichment vs. random. Researchers also found new candidate genes involved in cell adhesion and wound healing, co-expressed in the same skin layer, and implicated in EB comorbidities such as skin cancer.

Conclusion

Mavatar Discovery enabled researchers to build a disease-relevant pathway for EB despite limited direct data. By contextualizing known mutations within a larger biological framework, the platform revealed new mechanistic insights and potential therapeutic avenues. This accelerated translational understanding in one of medicine's most complex rare diseases.

About Mavatar Discovery

Mavatar Discovery is a self-service, cloud-based research platform for pharma, biotech, and academic teams. Unlike traditional tools, it uses fully data-driven, real-time analysis to model disease mechanisms and reveal new therapeutic opportunities.

- From raw RNA-seq to translational insight — no coding, no delays
- Accelerated hypothesis generation — turn complex data into testable ideas
- Uncover biomarkers and disease mechanisms faster
- Thousands of transcriptomes in one integrated, searchable platform
- A smarter starting point for next-generation therapies

Want access to Mavatar Discovery?

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Disclaimer:
This case study is a fictive example, created for illustrative purposes to demonstrate the capabilities of the Mavatar Discovery platform.

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