Improving targeted treatment opportunities of cancer patients via functional CRISPR-Select platform

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Kræftens Bekæmpelse

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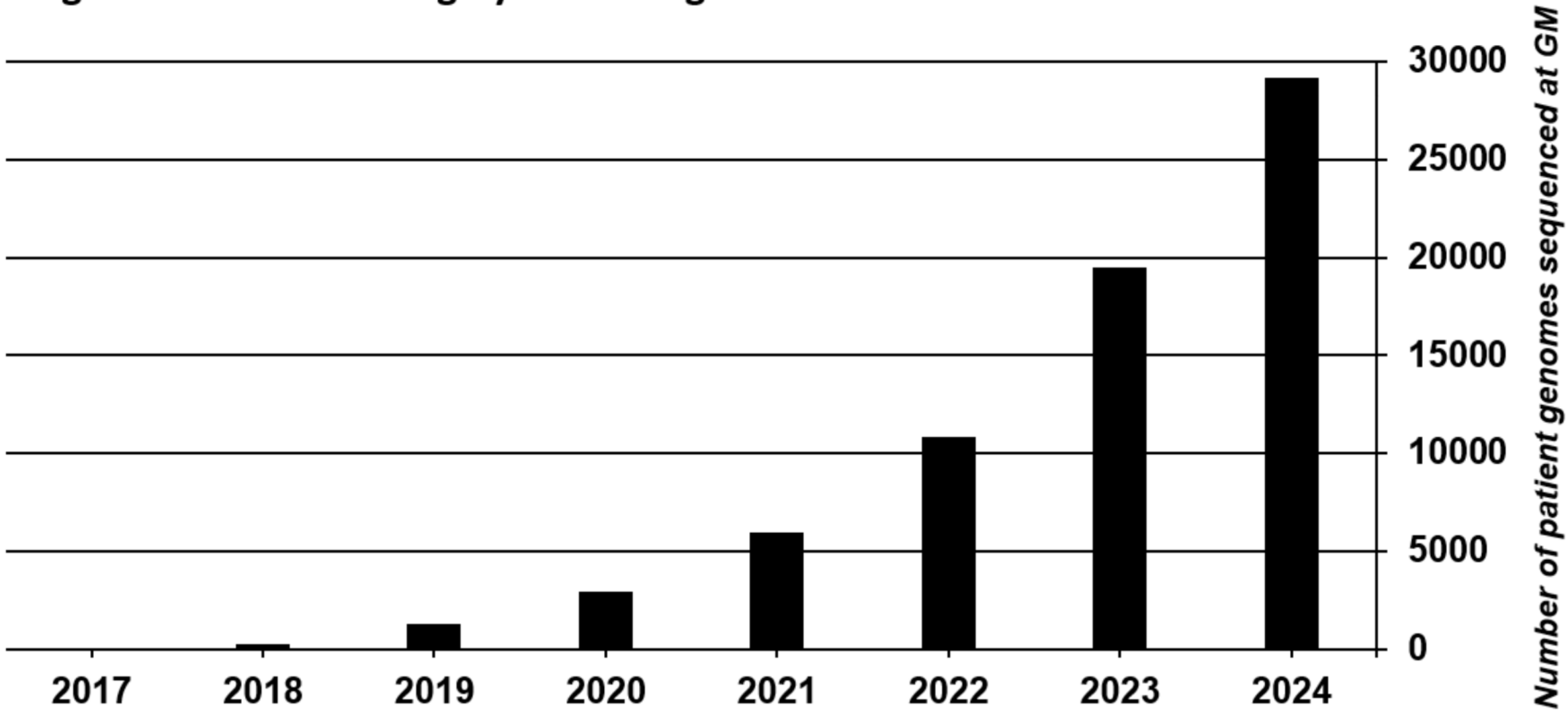
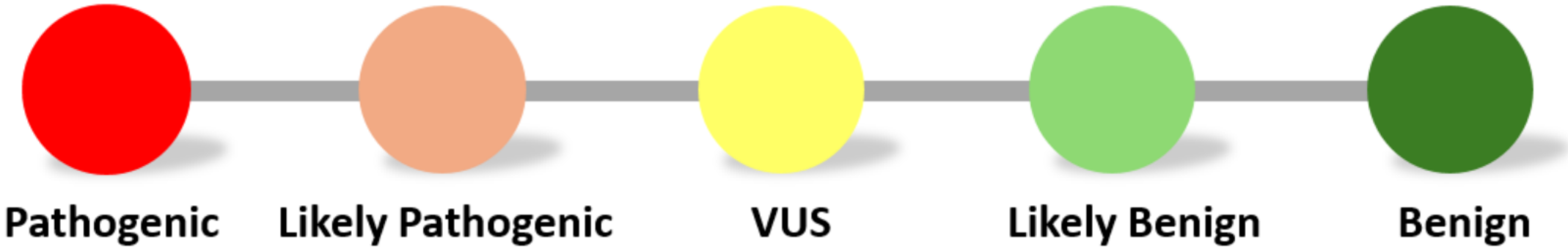
- **Our Department of Genomic Medicine (GM) at Rigshospitalet has sequenced close to 30,000 patient genomes.**
- **Each genome contains roughly 5 million genetic variants.**



Variant of Uncertain Significance (VUS) remains as a bottleneck in advancing Precision Medicine

A Spectrum of genetic variants identified in patients with varying clinical significance



VUS is the most frequently observed genetic variants among patients

Precision Medicine

Variant of Uncertain Significance (VUS) remains as a bottleneck in advancing Precision Medicine

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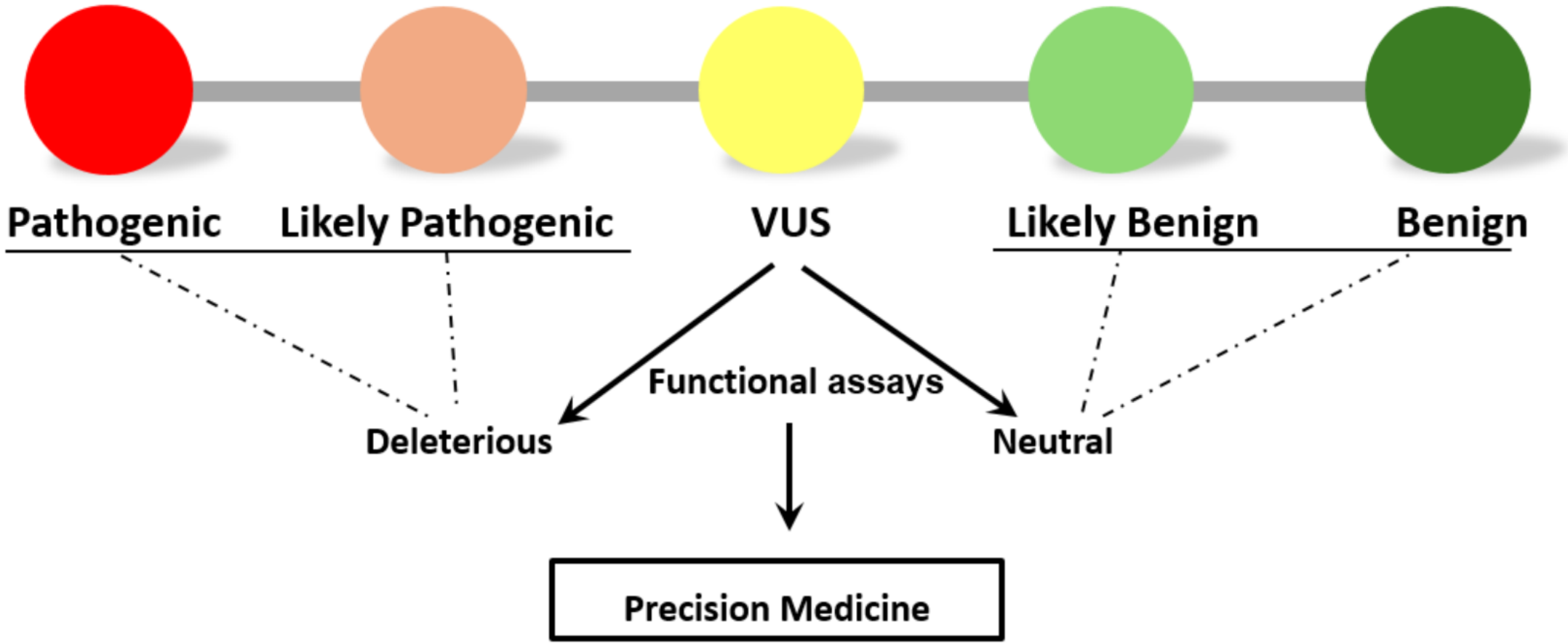
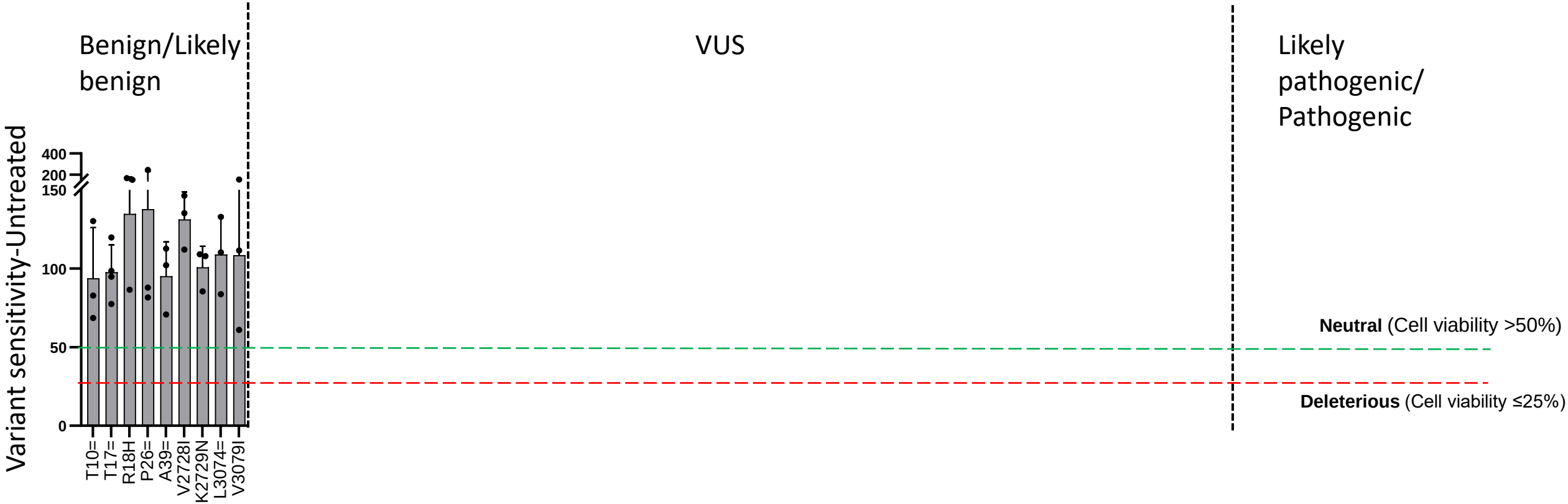


To address our aim, we used **CRISPR-Select** (PMID: 36471068) to determine the pathogenicity of VUS observed in *BRCA2. Please check poster #48 for more details on CRISPR-Select*

Clinical calibration of CRISPR-Select using *BRCA2*



Bose *et al.* J Clin Invest. 2025;135(12):e181879. doi:10.1172/JCI181879

Conclusions

- **CRISPR-Select** provides fast, scalable, and clinically actionable classification of *BRCA2* variants (see **Bose et al., JCI, 2025** — scan the barcode below).
- The CRISPR-Select assay demonstrated **100% sensitivity** and **100% specificity**.
- When combined with clinical guidelines, CRISPR-Select can optimize patient care and management while advancing precision medicine.
- **The Center for Genomic Medicine at Rigshospitalet is currently implementing clinical *BRCA2* functional variant classification.**
- Building on the *BRCA2* initiative, we are expanding this diagnostic service to additional clinically relevant genes (e.g., *BRCA1*).
- *Please check poster #48 for more details on our study and its clinical potential.*

Bose et al., JCI, 2025

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