

ORPHAN VASCULAR ANOMALIES: USING HPC MULTILAYER NETWORKS TO IDENTIFY THERAPEUTIC OPPORTUNITIES

MARENOSTRUM5 GPP

Led by



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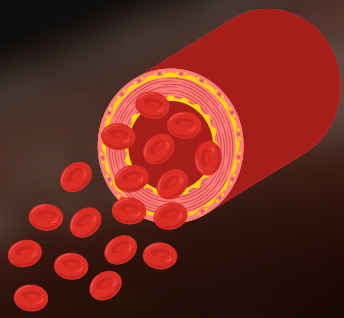


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1 WHAT ARE **VASCULAR ANOMALIES?**



Vascular anomalies are rare disorders that cause **an unusual growth of blood and lymph vessels** in any part of the body

Its severity depends on where the lesion is and its size, and it **can lead to severe complications**



These complications range from chronic pain and bleeding **to organ damage or even heart failure**

They are caused by genetic mutations, and current therapies can't ensure full efficacy or that they won't reappear



THE CHALLENGE: 2

THE RAREST ANOMALIES



Recently, they discovered that many mutations behind these anomalies **are the same that cause adult cancers**

This allows to **repurpose current oncology drugs** for certain patients with vascular anomalies



However, the rarest anomalies (**orphan anomalies**) can't be currently treated and **require a new research approach**

3 THE ROLE OF **HPC** RESOURCES



The team analysed **the DNA and gene activity of blood vessel cells** from patient lesions to find exploitable vulnerabilities in these diseases

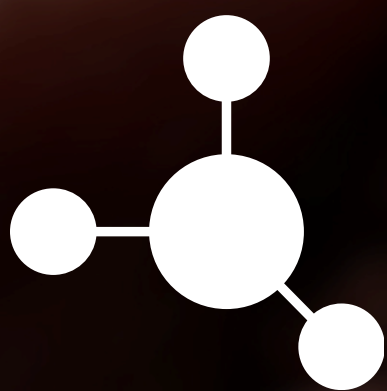
That means a huge amount of complex data that needs **powerful computational tools to be processed**



The team ran **a large-scale analysis** to compare genetic variants, biological pathways and gene activity patterns **across all patient samples**

REPURPOSING THERAPIES TO ORPHAN ANOMALIES

4



They discovered **molecular vulnerabilities** present not only in lesions with known mutations, but also **in these that the mutation cause is unknown**

These findings provide **an evidence-based framework** to help to repurpose current oncological drugs to **treat orphan vascular anomalies** that right now can't be treated



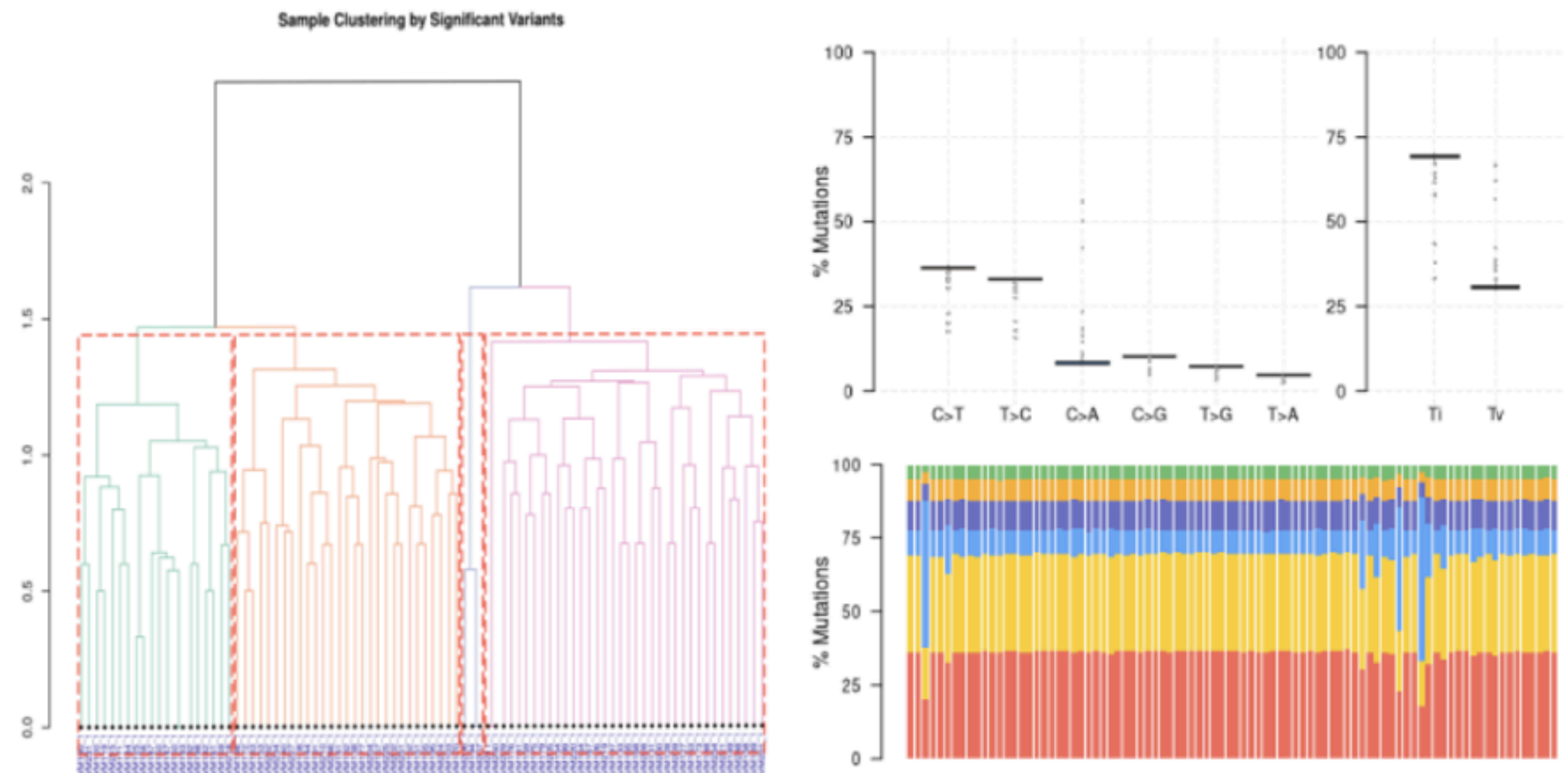


Figure: Genetic clustering groups individuals by DNA similarity to reveal population structures, while ti/tv analysis categorizes SNPs into transitions and transversions and visualizes their distributions.