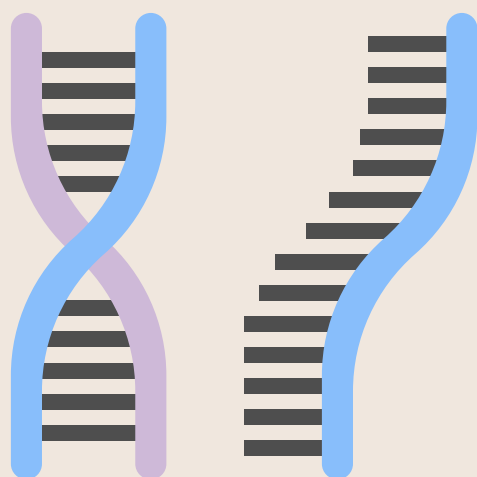


THE DARK GENOME AND RARE DISEASE



Current clinical diagnostic pathways focus on well-researched protein-coding regions, but any part of the genome can hold important information for rare disease diagnosis.

The relevance of noncoding variations and transcripts in the dark genome is becoming more apparent due to new 'omics technologies.

We now know that 95% of genetic variation can be found in the dark genome. Some of that variation is significant in the diagnosis of rare disease.

We also know that noncoding transcripts can be effectively used as biomarkers for rare disease diagnosis, because of their temporal, tissue, cell-type or disease-specificity.

HOW COULD THIS HELP?

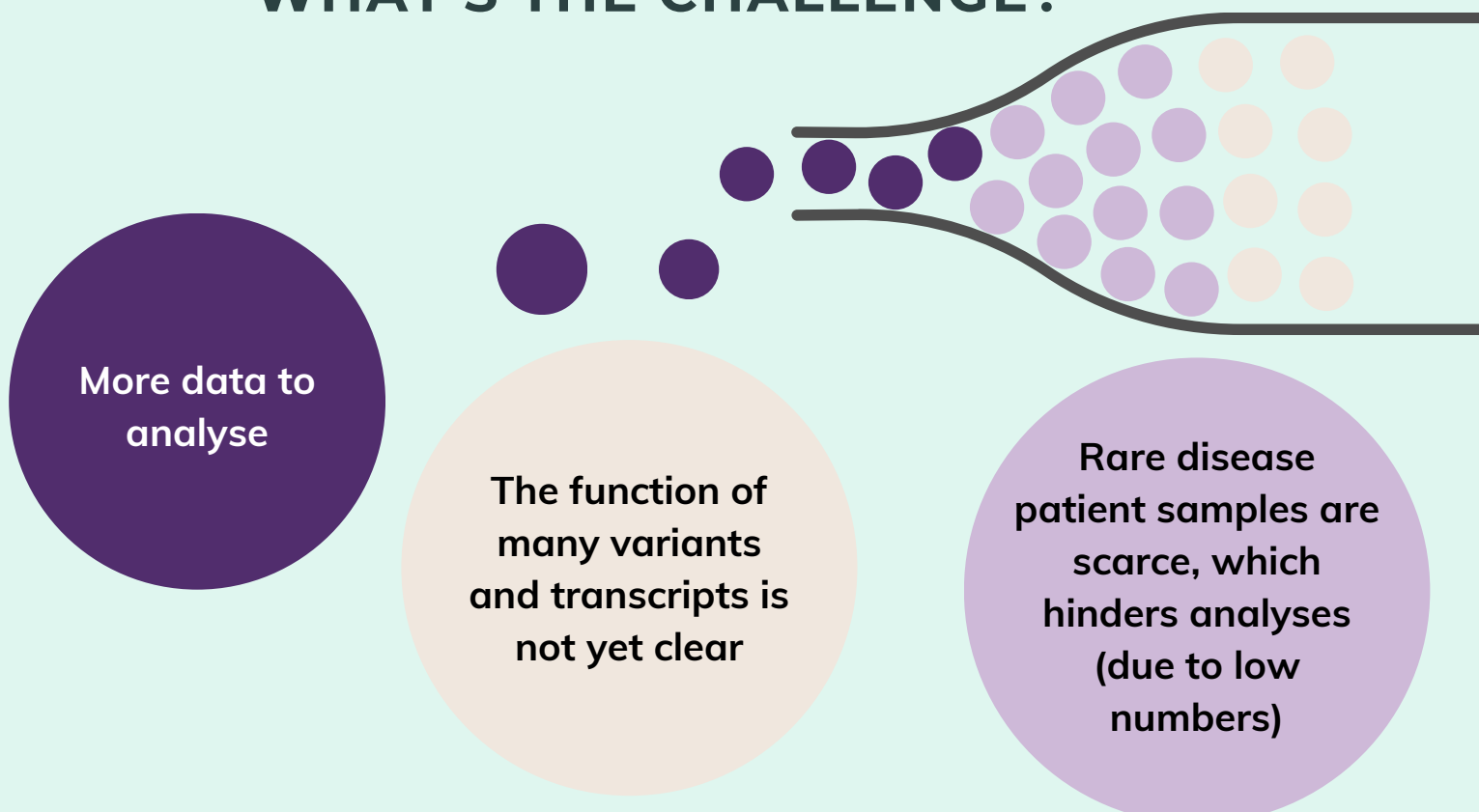


More patients getting diagnosed

More drug targets identified

Better outcomes from DNA and RNA samples collected

WHAT'S THE CHALLENGE?



TO BRIDGE THE DIAGNOSTIC GAP

- ✓ Standardise collection of the 'right' data to support noncoding analysis
- ✓ Create standardised data analysis pipelines with noncoding specific databases
- ✓ Prepare guidelines for interpreting noncoding results
- ✓ Scale-up data processing and storage systems to handle increased data analysis demands



The current landscape supports a greater use of noncoding genome-based diagnoses in clinical care as a complement to traditional protein-coding region-based approaches