

POPULATION LEVEL SCREENING OF PID PATIENTS BY GRID - GENOMICS OF RARE IMMUNE DISORDERS - GENE PANEL

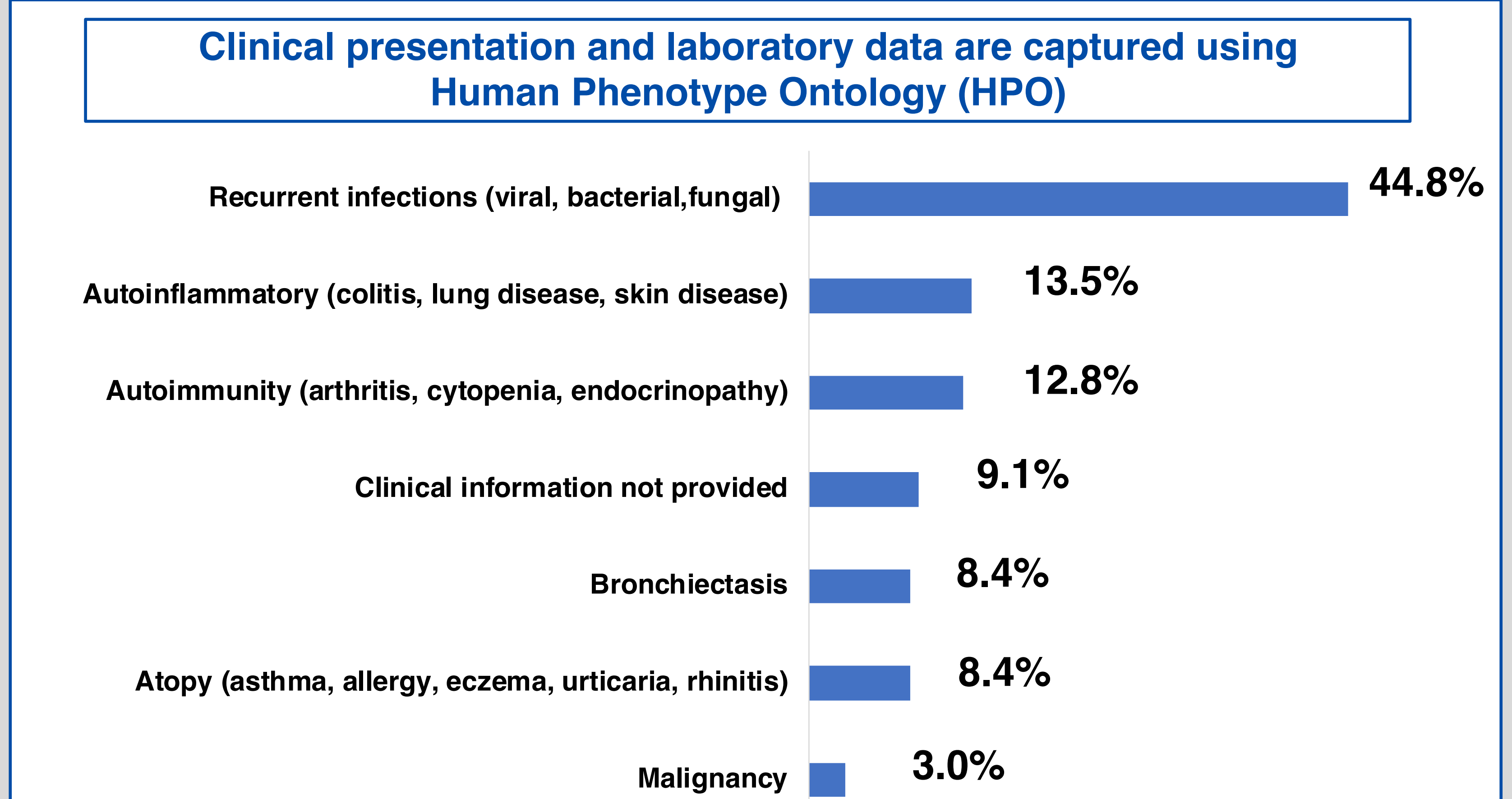
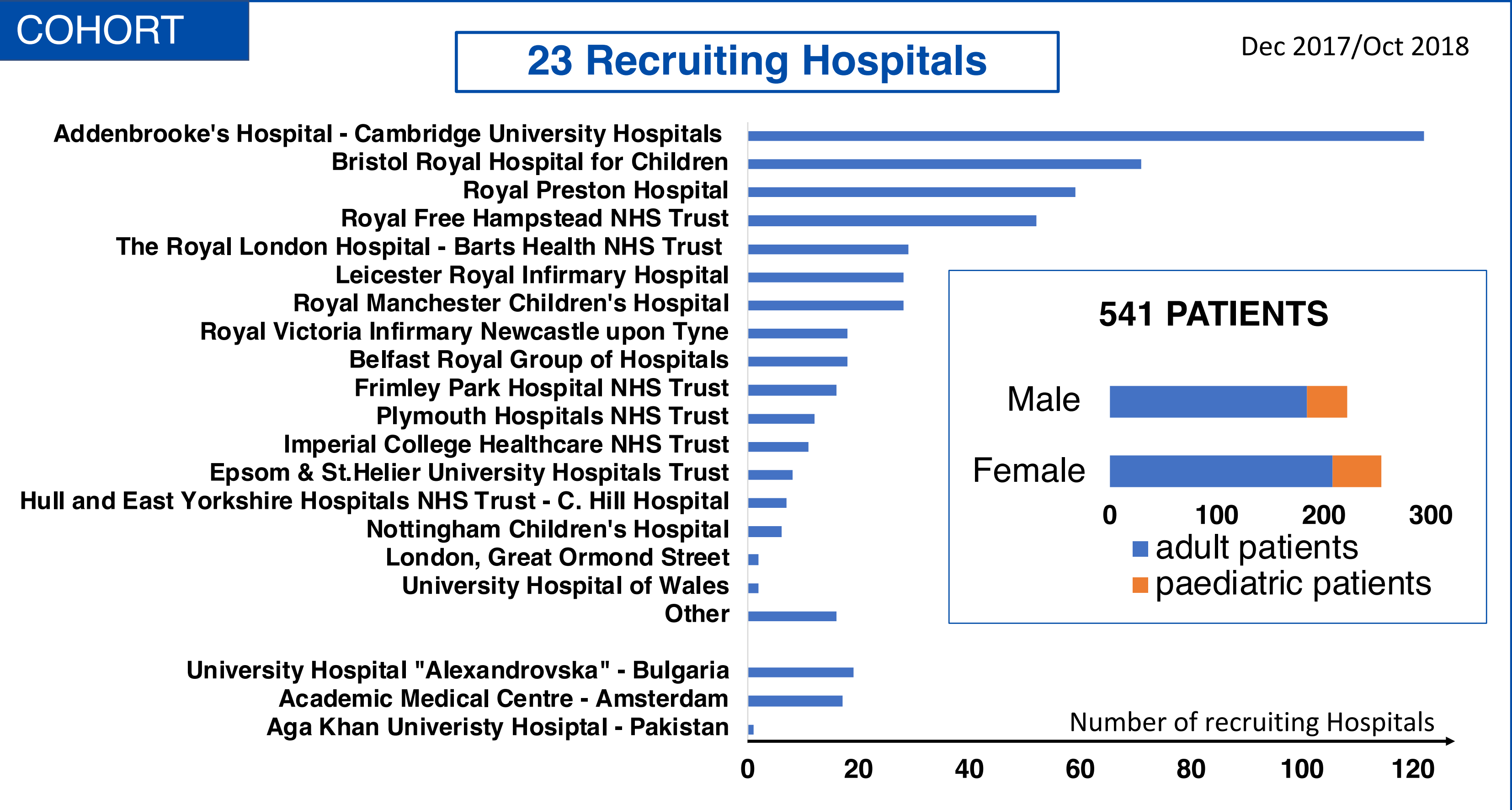


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SUMMARY A conclusive molecular diagnosis for patients with Primary Immune Disorders (PID) is often challenging due to the high number of genes containing causal variants and the heterogeneity of clinical presentations. A targeted high throughput sequencing gene panel covering all known PID genes can be an invaluable diagnostic tool.



VARIANT PRIORITISATION AND INTERPRETATION

A COMPREHENSIVE GENE LIST including the 2015 IUIS genes

(Version GRID 2.0) www.gridgenomics.org.uk

279 Tier 1 genes

IV. Diseases of Immune dysregulation	V. Congenital defects of phagocyte number, function, or both	VI. Defects in intrinsic and innate immunity	VII. Auto-inflammatory disorders	VIII. Complement deficiencies	IX. Phenotypes of PID
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