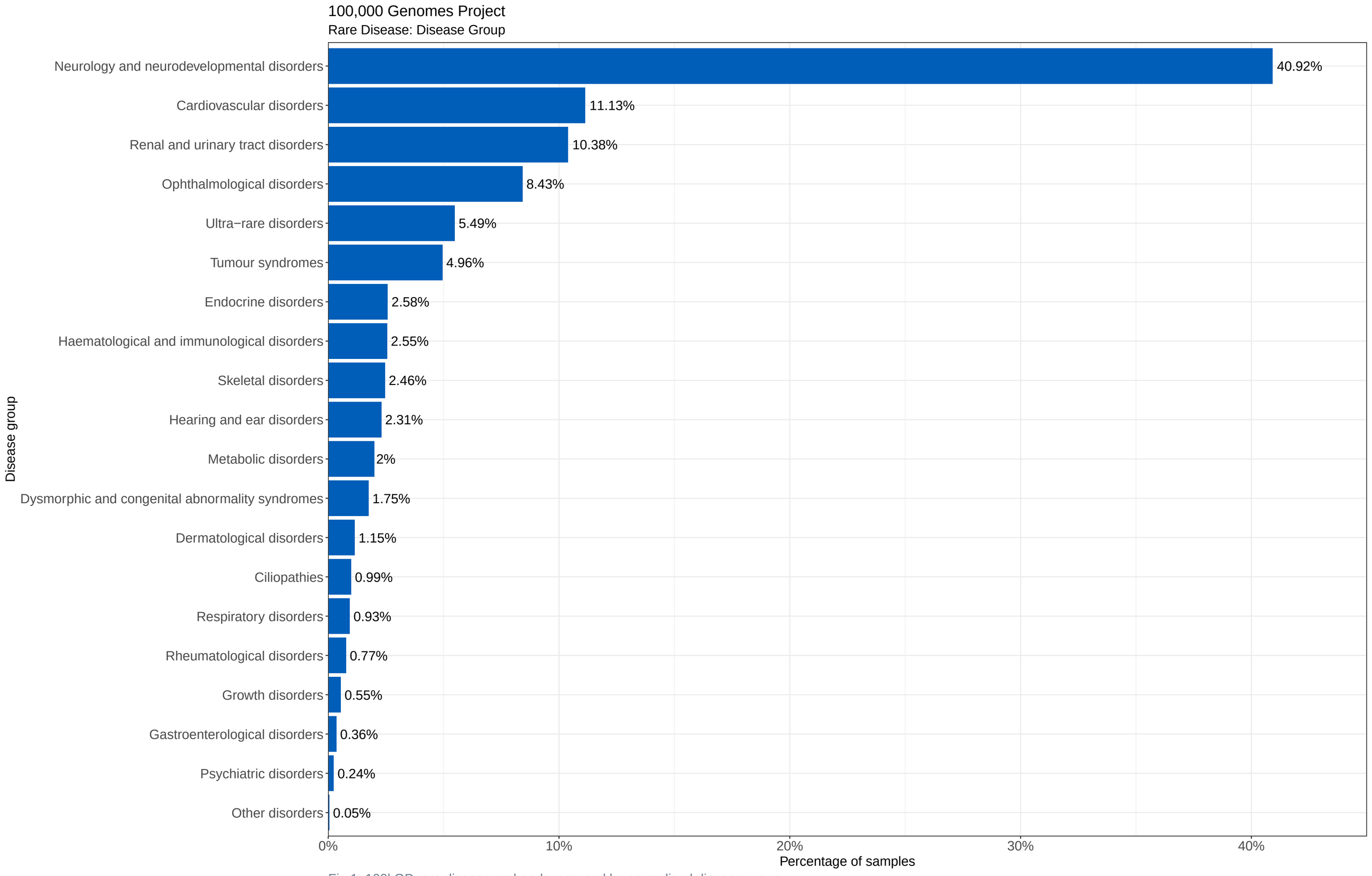

Fig 1. 100kGP rare disease probands, grouped by normalised disease group.

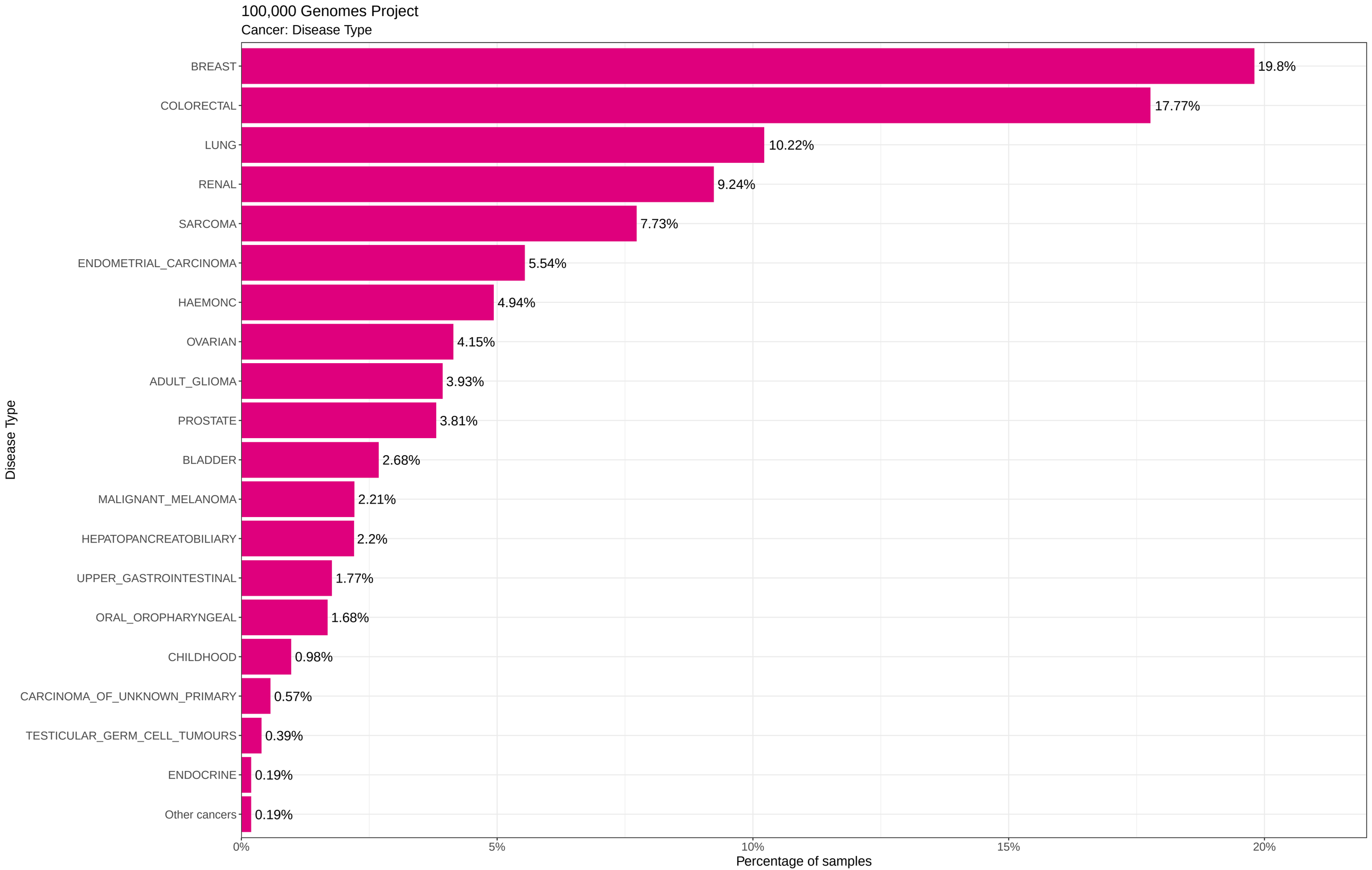

Fig 2. 100kGP cancer participants, grouped by disease type.

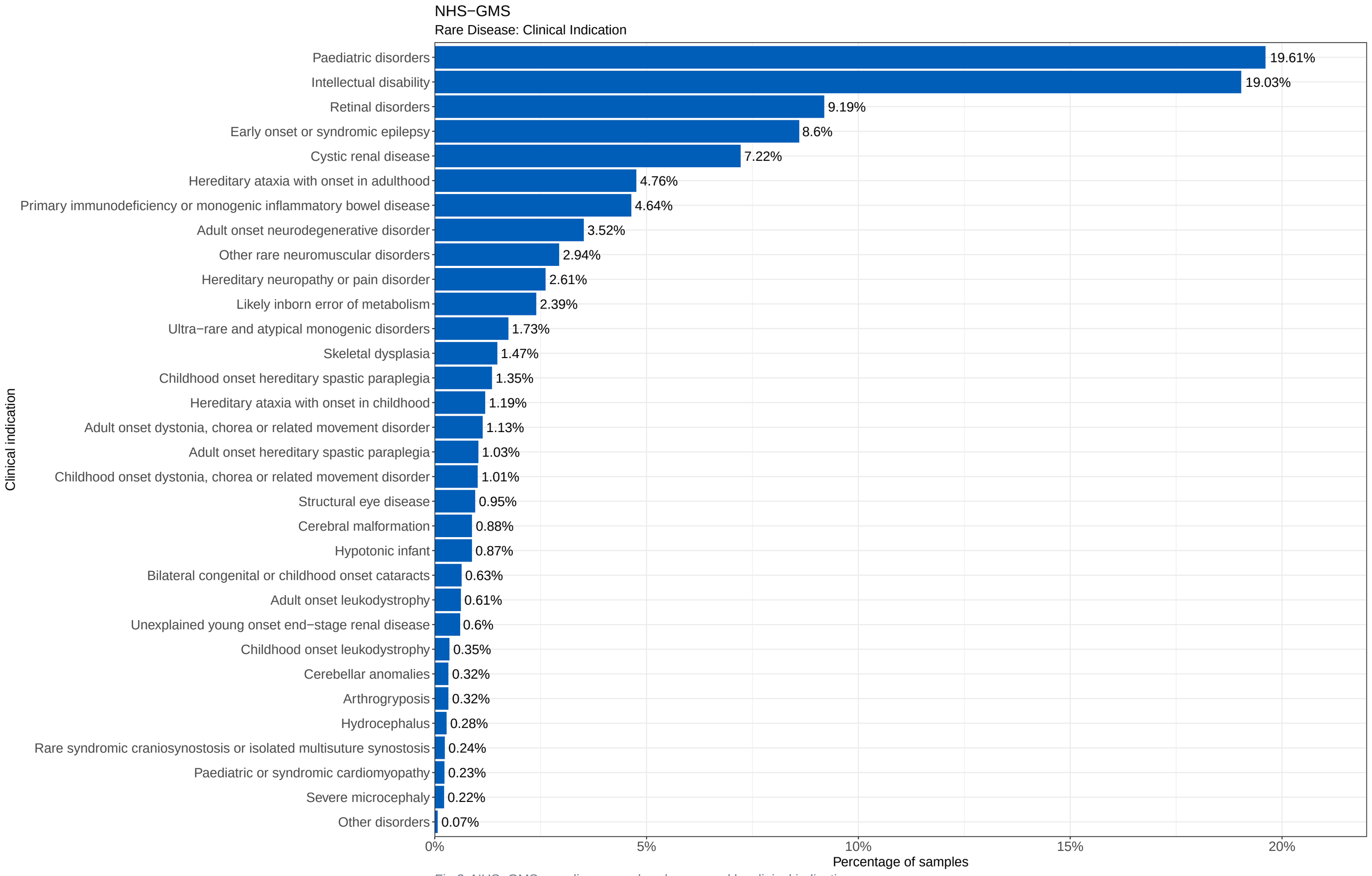

Fig 3. NHS−GMS rare disease probands, grouped by clinical indication.

NHS−GMS

Cancer: Clinical Indication

Clinical indication

Acute Lymphoblastic Leukaemia 18.54%
Acute Myeloid Leukaemia 11.83%
Paediatric Tumours 5.66%
Glioblastoma − Adult 4.9%
Solid Tumour Exhausted all Standards of Care Testing and Treatment− Adult 4.66%
Spindle Cell Soft Tissue Tumour Differential 4.23%
High Grade Ovarian Carcinoma (WGS PILOT) 3.42%
High Grade Glioma − Adult 3.18%
Neuroblastoma − Paediatric 2.8%
Osteosarcoma 2.71%
Wilms Tumours − Paediatric 2.42%
Ewing Sarcoma of Bone 1.52%
Pilocytic Astrocytoma − Paediatric 1.43%
Meningioma − Adult 1.28%
Glioma − Adult 1.28%
Undifferentiated tumour 1.24%
Astrocytoma − Adult 1.24%
Alveolar Rhabdomyosarcoma 1.24%
Proven or Suspected Haematological Tumours Exhausted all Standard of Care Testing and Treatment 1.09%
High Grade Glioma − Paediatric 1%
Other cancers 24.33%

0% 10% 20% 30%

Percentage of samples

Fig 4. NHS−GMS cancer participants, grouped by clinical indication.