Supplementary Fig.1 Number of variants per clinical significance

a In the Clinical significance box, the number of variants per clinical significance is displayed for 1,052,579 ClinVar variants in TogoVar. Note that the number is counted for each significance when two or more clinical significances are assigned to one variant.

b The number per clinical significance for 123,160 variants included in the five datasets of JPN UNION is displayed.

b

a

| Statistics / Filters | Reset |
|--|-----------|
| Dataset | ^ |
| ☐ All | 1,052,579 |
| ☐ S WGS GEM-J WGA | 138,718 |
| ☐ SWES JGA NGS | 56,079 |
| ☐ SNP JGA SNP | 19,190 |
| ☐ S WGS ToMMo 8.3KJPN | 146,704 |
| ☐ S WES HGVD | 51,670 |
| gnomAD genomes | 422,465 |
| gnomAD exomes | 521,805 |
| ✓ Sease ClinVar | 1,052,579 |
| Alternative allele frequency | ~ |
| Variant calling quality | ~ |
| Variant type | ~ |
| Clinical significance | ^ |
| ✓ All | 1,052,579 |
| ✓ Not in ClinVar | 0 |
| Pathogenic | 154,665 |
| Likely pathogenic | 77,153 |
| Uncertain significance | 601,500 |
| Likely benign | 404,249 |
| B Benign | 206,181 |
| Conflicting interpretations of pathogenicity | |
| . • • | 18,709 |
| ✓ DR Drug response | 2,228 |
| Association | 365 |
| Risk factor | 771 |
| Protective | 74 |
| ✓ AF Affects | 192 |
| Other | 1,213 |
| Not provided | 17,833 |
| Association_not found | 2 |
| Consequence | |

Statistics / Filters Reset **Dataset** ☐ All 123,160 **GEM-J WGA** 101,183 WGS **JGA NGS** 31,297 WES SNP **JGA SNP** 12,964 104,709 ToMMo 8.3KJPN WGS HGVD WES 47,402 94,795 🔲 🥮 WGS gnomAD genomes 87,336 ■ WES gnomAD exomes ClinVar 123,160 Disease Alternative allele frequency Variant calling quality Variant type Clinical significance 123,160 Not in ClinVar 0 Pathogenic 5,466 Likely pathogenic 2,519 ✓ (us) Uncertain significance 56,789 Likely benign 49,484 **✓** (B) Benign 92,481 ✓ (c) Conflicting interpretations of pathogenicity 3,159 **✓** (DR) Drug response 446 ✓ A Association 176 Risk factor 235 ✓ (PR) Protective 43

49

262

1,515

2

AF Affects

Consequence

Other

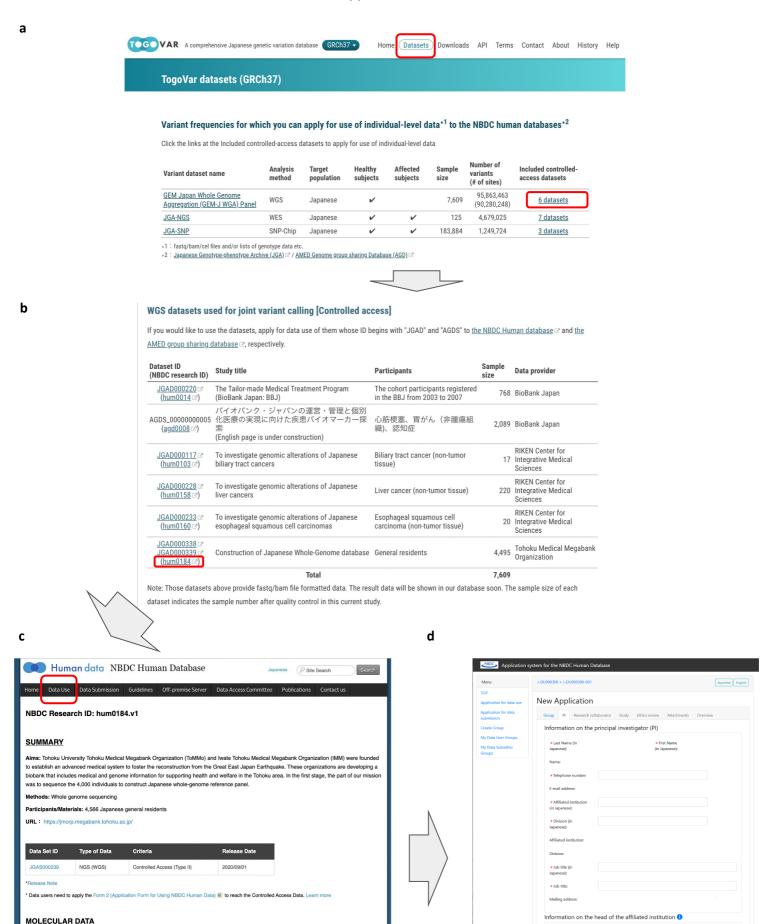
NP Not provided

Association_not found

(0)

Supplementary Fig.2 How to access individual-level genome data in JGA

a Identify the allele frequency dataset (GEM-J WGA) that contains the variant of interest. **b** Focus on hum0184 among the six individual-level genomic datasets of GEM-J WGA. **c** Obtain an overview of the hum0184 dataset in the NBDC human database. d Submit a data-use application.



4,566 Japanese general residents

Illumina [HiSeq 2500, NovaSeq 6000]

WGS

Target Loci for Capture Methods