

## **Description of Additional Supplementary Files**

File Name: Supplementary Data 1

Description: Summary of sample information and sequencing statistics

File Name: Supplementary Data 2

Description: . Summary of clinical traits

File Name: Supplementary Data 3

Description: SV statistics of different filtering processes for each sample

File Name: Supplementary Data 4

Description: Manually checked INVs of HG002

File Name: Supplementary Data 5

Description: Summary of Uniquely detected SVs by one caller and the overlaps with benchmark set

File Name: Supplementary Data 6

Description: Annotation of repeat sequences in SVs

File Name: Supplementary Data 7

Description: Summary of PCR validation results of singletons

File Name: Supplementary Data 8

Description: Summary of PCR validation results of novel non-singletons

File Name: Supplementary Data 9

Description: SVs and associated genes in GWAS, OMIM and COSMIC datasets

File Name: Supplementary Data 10

Description: Association for SVs and clinical phenotypes

File Name: Supplementary Data 11

Description: Information of SVs with significant PBS between Southern and Northern Chinese