Table E3: **Comparison of causal genetic variant detection between conventional methods and the customized GSA**

Item	**Conventional Methods(Numbers)**		**GSA technology (Numbers)**
Method	**Previously detected variants**	**Known variants that could be investigated by GSA**	**Replicated variants by GSA**
SNV	71 *	37	30
CNV	4	4	3
Method	**Newly found variants replicated by Sanger sequencing**	**Newly found variants that could be investigated by Sanger sequencing**	**Newly found variants by GSA**
SNV	38	41	46**
CNV	none	none	2

* 34 out of 71 variants did not include when the GSA was designed.

** 5 out of 46 variants could not be investigated (lack of DNA).

GSA: Global Screening Array, CNV: copy number variants, SNV: single nucleotide variants