Biomarker	Chr	Start	End	Beta	SE	P Value
GPNMB	2	89,613,000	89,613,200	-9.91E-01	1.56E-01	3.33E-10
PD-L2	3	98,411,600	98,411,800	4.09E-01	4.47E-02	3.92E-19
IL-18	5	70,393,100	70,393,300	4.18E-01	5.61E-02	2.38E-13
ST1A1	16	28,613,645	28,613,845	7.86E-01	6.93E-02	1.61E-27
hOSCAR	19	54,558,900	54,559,100	-7.21E-01	6.27E-02	1.61E-28
CD48	1	158,867,600	158,867,800	-2.82E-01	4.71E-02	3.15E-09
FCRLB	1	161,640,580	161,640,780	6.17E-01	9.11E-02	2.42E-11
LY9	1	179,455,600	179,455,800	5.36E-01	8.86E-02	2.17E-09
ICAM-2	3	98,410,600	98,410,800	6.48E-01	3.95E-02	5.67E-53
Siglec-9	3	98,411,800	98,413,400	6.82E-01	4.17E-02	3.48E-52
CD200R1	3	98,411,800	98,413,400	5.08E-01	4.45E-02	3.71E-28
VEGFR-3	3	98,414,600	98,414,800	5.96E-01	4.17E-02	1.10E-41
ICAM-3	3	98,899,900	98,900,100	3.96E-01	5.52E-02	1.45E-12
AMBP	5	745,070	745,270	-2.84E-01	4.51E-02	5.05E-10
MIC-AB	6	32,496,600	32,496,800	6.19E-01	6.69E-02	3.35E-19
CCL19	6	32,522,200	32,522,400	-4.67E-01	5.37E-02	1.93E-17
FR-gamma	11	63,443,100	63,445,300	-1.03E+00	9.83E-02	5.17E-24
FR-gamma	11	67,331,355	67,331,955	-1.23E+00	1.03E-01	1.64E-30
CNTN1	12	45,909,600	45,909,800	-2.85E-01	4.69E-02	1.73E-09
CCL4	17	36,392,670	36,394,670	1.45E-01	2.29E-02	4.65E-10
CCL15	17	39,210,800	39,211,000	-9.21E-01	1.23E-01	1.45E-13
SMPD1	19	35,863,600	35,863,800	3.69E-01	6.08E-02	1.97E-09
MIA	19	41,381,925	41,385,125	-1.23E+00	1.69E-01	8.58E-13
hK11	19	51,508,940	51,510,740	1.41E+00	2.22E-01	3.84E-10
WFDC2	20	44,204,435	44,205,035	4.17E-01	6.81E-02	1.37E-09

Table 4.1: CN-GWAS results. The CNVs in the top section were used to select the 15 individuals for resequencing.

4.3 Results and Discussion

Overall, 872 individuals had both genotyping and protein data which passed QC. CNVnator reported 243,987 CNVs, which after post-processing resulted in 23,831 variants to be included in our analysis. We found 30 CNVs to be associated with 17 biomarkers (table 4.1).

The quality of our long-read sequencing results was mixed. While most samples received high coverage, in three cases less than half of all ZMWs produced high-quality reads. The CNV on chromosome 2 was not called by any of our long-read callers. The CNV on chromosome 3 was only detected by SVIM, which also called all copy numbers in accordance with CNVnator. The CNV on chromosome 5 was not detected in the long-read data. The coverage in this region was spotty at best, in both Illumina and