Table S9. Performance of 20 pipelines on NA24385 and NA24631 data sets using both reference genomes GRCh38 and GRCh37 for SNVs and InDels

	GRCh37													
	NA24385													
	SNV							InDel						
Pipeline	TP	FP	FN	Sensitivity	Precision	F-Score	FDR	TP	FP	FN	Sensitivity	Precision	F-Score	FDR
Bowtie_DeepVariant	39912	663	1005	0.975	0.984	0.980	0.016	3066	254	188	0.942	0.923	0.933	0.077
Bowtie_FreeBayes	40001	574	970	0.976	0.986	0.981	0.014	2791	529	261	0.914	0.841	0.876	0.159
Bowtie_GATK	39901	674	986	0.976	0.983	0.980	0.017	3089	231	168	0.948	0.930	0.939	0.070
Bowtie_SAMtools	39967	608	993	0.976	0.985	0.980	0.015	2591	729	308	0.894	0.780	0.833	0.220
BWA_DeepVariant	40105	470	857	0.979	0.988	0.984	0.012	3145	175	121	0.963	0.947	0.955	0.053
BWA_FreeBayes	39537	1038	1837	0.956	0.974	0.965	0.026	3013	307	182	0.943	0.908	0.925	0.092
BWA_GATK	40089	486	897	0.978	0.988	0.983	0.012	3157	163	127	0.961	0.951	0.956	0.049
BWA_SAMtools	40091	484	911	0.978	0.988	0.983	0.012	2607	713	362	0.878	0.785	0.829	0.215
MOSAIK_DeepVariant	39909	666	1002	0.976	0.984	0.980	0.016	3061	259	179	0.945	0.922	0.933	0.078
MOSAIK_FreeBayes	39885	690	1871	0.955	0.983	0.969	0.017	2997	323	190	0.940	0.903	0.921	0.097
MOSAIK_GATK	7979	32596	158	0.981	0.197	0.328	0.803	1501	1819	1820	0.452	0.452	0.452	0.548
MOSAIK_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Novoalign_DeepVariant	40098	477	863	0.979	0.988	0.984	0.012	3189	131	103	0.969	0.961	0.965	0.039
Novoalign_FreeBayes	39881	694	1046	0.974	0.983	0.979	0.017	2984	336	205	0.936	0.899	0.917	0.101
Novoalign_GATK	40071	504	904	0.978	0.988	0.983	0.012	3183	137	111	0.966	0.959	0.963	0.041
Novoalign_SAMtools	40093	482	893	0.978	0.988	0.983	0.012	2655	665	279	0.905	0.800	0.849	0.200
SOAP_DeepVariant	-	-	-	-	-	-	-	-	-	-	-	-	-	-
SOAP_FreeBayes	39742	833	1806	0.957	0.979	0.968	0.021	1530	1790	1539	0.499	0.461	0.479	0.539
SOAP_GATK	39511	1064	1901	0.954	0.974	0.964	0.026	1518	1802	1639	0.481	0.457	0.469	0.543
SOAP_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-

GRCh38													
NA24385													
	SNV							**InDel**					
Pipeline	**TP**	**FP**	**FN**	**Sensitivity**	**Precision**	**F-Score**	**FDR**	**TP**	**FP**	**FN**	**Sensitivity**	**Precision**	**F-Score**	**FDR**
Bowtie_DeepVariant	41071	1769	1099	0.974	0.959	0.966	0.041	4028	484	365	0.917	0.893	0.905	0.107
Bowtie_FreeBayes	40921	1919	1231	0.971	0.955	0.963	0.045	3955	557	401	0.908	0.877	0.892	0.123
Bowtie_GATK	41090	1750	1148	0.973	0.959	0.966	0.041	4021	491	312	0.928	0.891	0.909	0.109
Bowtie_SAMtools	42103	737	1071	0.975	0.983	0.979	0.017	3882	630	476	0.891	0.860	0.875	0.140
BWA_DeepVariant	42267	573	1031	0.976	0.987	0.981	0.013	4101	411	261	0.940	0.909	0.924	0.091
BWA_FreeBayes	42063	777	1261	0.971	0.982	0.976	0.018	4001	511	371	0.915	0.887	0.901	0.113
BWA_GATK	42152	688	1081	0.975	0.984	0.979	0.016	4095	417	278	0.936	0.908	0.922	0.092
BWA_SAMtools	42257	583	1049	0.976	0.986	0.981	0.014	3861	651	498	0.886	0.856	0.870	0.144
MOSAIK_DeepVariant	41082	1758	1111	0.974	0.959	0.966	0.041	4011	501	325	0.925	0.889	0.907	0.111
MOSAIK_FreeBayes	42077	763	1293	0.970	0.982	0.976	0.018	3967	545	355	0.918	0.879	0.898	0.121
MOSAIK_GATK	13830	29010	941	0.936	0.323	0.480	0.677	3561	951	2094	0.630	0.789	0.701	0.211
MOSAIK_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Novoalign_DeepVariant	42274	566	1023	0.976	0.987	0.982	0.013	4131	381	234	0.946	0.916	0.931	0.084
Novoalign_FreeBayes	42051	789	1290	0.970	0.982	0.976	0.018	3951	561	392	0.910	0.876	0.892	0.124
Novoalign_GATK	42139	701	1079	0.975	0.984	0.979	0.016	4129	383	241	0.945	0.915	0.930	0.085
Novoalign_SAMtools	42251	589	1059	0.976	0.986	0.981	0.014	3897	615	420	0.903	0.864	0.883	0.136
SOAP_DeepVariant	-	-	-	-	-	-	-	-	-	-	-	-	-	-
SOAP_FreeBayes	40123	2717	2124	0.950	0.937	0.943	0.063	3592	920	2042	0.638	0.796	0.708	0.204
SOAP_GATK	40891	1949	2041	0.952	0.955	0.953	0.045	3402	1110	2104	0.618	0.754	0.679	0.246
SOAP_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-
GRCh37														
NA24631														
	SNV							**InDel**					
Pipeline	**TP**	**FP**	**FN**	**Sensitivity**	**Precision**	**F-Score**	**FDR**	**TP**	**FP**	**FN**	**Sensitivity**	**Precision**	**F-Score**	**FDR**
Bowtie_DeepVariant	40482	649	992	0.976	0.984	0.980	0.016	2783	207	197	0.934	0.931	0.932	0.069
Bowtie_FreeBayes	40412	719	882	0.979	0.983	0.981	0.017	2421	569	280	0.896	0.810	0.851	0.190
Bowtie_GATK	40241	890	1063	0.974	0.978	0.976	0.022	2542	448	214	0.922	0.850	0.885	0.150

Pipeline	TP	FP	FN	Sensitivity	Precision	F-Score	FDR	TP	FP	FN	Sensitivity	Precision	F-Score	FDR
Bowtie_SAMtools	40462	669	1003	0.976	0.984	0.980	0.016	2391	599	318	0.883	0.800	0.839	0.200
BWA_DeepVariant	40580	551	748	0.982	0.987	0.984	0.013	2883	107	142	0.953	0.964	0.959	0.036
BWA_FreeBayes	40089	1042	2833	0.934	0.975	0.954	0.025	2481	509	259	0.905	0.830	0.866	0.170
BWA_GATK	40543	588	781	0.981	0.986	0.983	0.014	2858	132	181	0.940	0.956	0.948	0.044
BWA_SAMtools	40559	572	751	0.982	0.986	0.984	0.014	2309	681	1078	0.682	0.772	0.724	0.228
MOSAIK_DeepVariant	40213	918	1062	0.974	0.978	0.976	0.022	2691	299	203	0.930	0.900	0.915	0.100
MOSAIK_FreeBayes	40132	999	2041	0.952	0.976	0.964	0.024	2313	677	294	0.887	0.774	0.827	0.226
MOSAIK_GATK	8320	32811	430	0.951	0.202	0.334	0.798	2013	977	924	0.685	0.673	0.679	0.327
MOSAIK_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Novoalign_DeepVariant	40583	548	734	0.982	0.987	0.984	0.013	2881	109	159	0.948	0.964	0.956	0.036
Novoalign_FreeBayes	40092	1039	2174	0.949	0.975	0.961	0.025	2311	679	274	0.894	0.773	0.829	0.227
Novoalign_GATK	40537	594	821	0.980	0.986	0.983	0.014	2866	124	179	0.941	0.959	0.950	0.041
Novoalign_SAMtools	40563	568	739	0.982	0.986	0.984	0.014	2351	639	351	0.870	0.786	0.826	0.214
SOAP_DeepVariant	-	-	-	-	-	-	-	-	-	-	-	-	-	-
SOAP_FreeBayes	40051	1080	2641	0.938	0.974	0.956	0.026	2308	682	1031	0.691	0.772	0.729	0.228
SOAP_GATK	40031	1100	3021	0.930	0.973	0.951	0.027	2129	861	953	0.691	0.712	0.701	0.288
SOAP_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-
GRCh38														
NA24631														
	SNV							InDel						
Pipeline	TP	FP	FN	Sensitivity	Precision	F-Score	FDR	TP	FP	FN	Sensitivity	Precision	F-Score	FDR
Bowtie_DeepVariant	43492	837	1850	0.959	0.981	0.970	0.019	3412	227	562	0.859	0.938	0.896	0.062
Bowtie_FreeBayes	43351	978	1946	0.957	0.978	0.967	0.022	3333	306	653	0.836	0.916	0.874	0.084
Bowtie_GATK	43539	790	1831	0.960	0.982	0.971	0.018	3409	230	582	0.854	0.937	0.894	0.063
Bowtie_SAMtools	43678	651	1521	0.966	0.985	0.976	0.015	3278	361	780	0.808	0.901	0.852	0.099
BWA_DeepVariant	43918	411	1345	0.970	0.991	0.980	0.009	3467	172	477	0.879	0.953	0.914	0.047
BWA_FreeBayes	43609	720	1591	0.965	0.984	0.974	0.016	3389	250	603	0.849	0.931	0.888	0.069
BWA_GATK	43852	477	1591	0.965	0.989	0.977	0.011	3471	168	501	0.874	0.954	0.912	0.046
BWA_SAMtools	43893	436	1385	0.969	0.990	0.980	0.010	3219	420	809	0.799	0.885	0.840	0.115
MOSAIK_DeepVariant	43502	827	1820	0.960	0.981	0.970	0.019	3381	258	581	0.853	0.929	0.890	0.071
MOSAIK_FreeBayes	43590	739	1630	0.964	0.983	0.974	0.017	3331	308	621	0.843	0.915	0.878	0.085

MOSAIK_GATK	19436	24893	1023	0.950	0.438	0.600	0.562	3131	508	2601	0.546	0.860	0.668	0.140
MOSAIK_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-
Novoalign_DeepVariant	43921	408	1301	0.971	0.991	0.981	0.009	3482	157	471	0.881	0.957	0.917	0.043
Novoalign_FreeBayes	43552	777	1721	0.962	0.982	0.972	0.018	3311	328	684	0.829	0.910	0.867	0.090
Novoalign_GATK	43841	488	1610	0.965	0.989	0.977	0.011	3475	164	489	0.877	0.955	0.914	0.045
Novoalign_SAMtools	43901	428	1399	0.969	0.990	0.980	0.010	3299	340	724	0.820	0.907	0.861	0.093
SOAP_DeepVariant	-	-	-	-	-	-	-	-	-	-	-	-	-	-
SOAP_FreeBayes	42871	1458	2895	0.937	0.967	0.952	0.033	3201	438	2576	0.554	0.880	0.680	0.120
SOAP_GATK	43172	1157	2751	0.940	0.974	0.957	0.026	3099	540	2555	0.548	0.852	0.667	0.148
SOAP_SAMtools	-	-	-	-	-	-	-	-	-	-	-	-	-	-