Supplementary Materials

Figure S1. Bioinformatic pipeline for short-read WES and WGS data processing and variant calling of human mitogenomes.

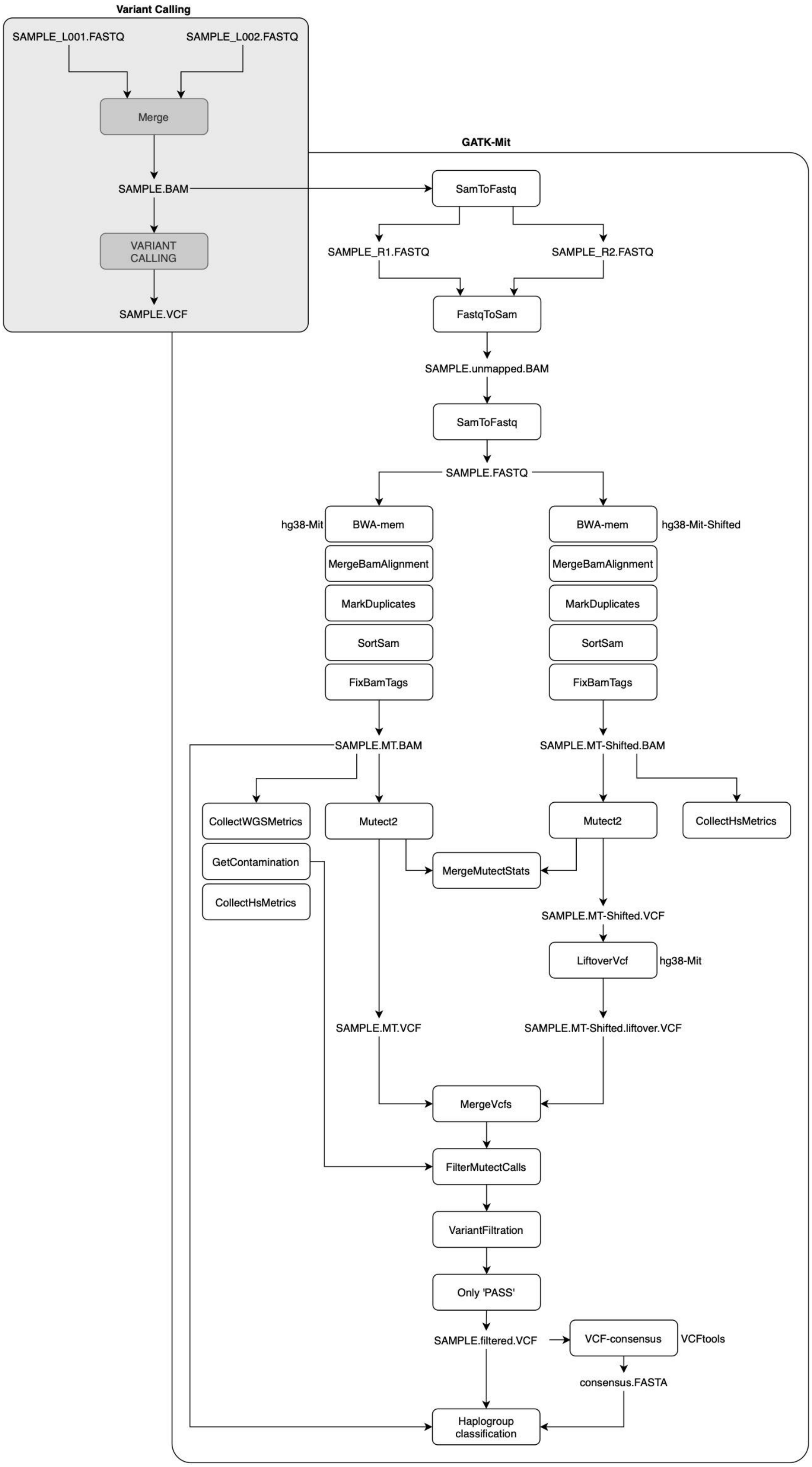

Figure S2. Bioinformatic pipeline for processing the long-read ONT data for human mitogenome reconstruction, including the four strategies used in the study: *de novo* assembly using long reads (in green), the hybrid *de novo* assembly using short and long reads (in red), the reference-based assembly using long reads (in blue), and the variant-calling strategy using long reads (in purple).

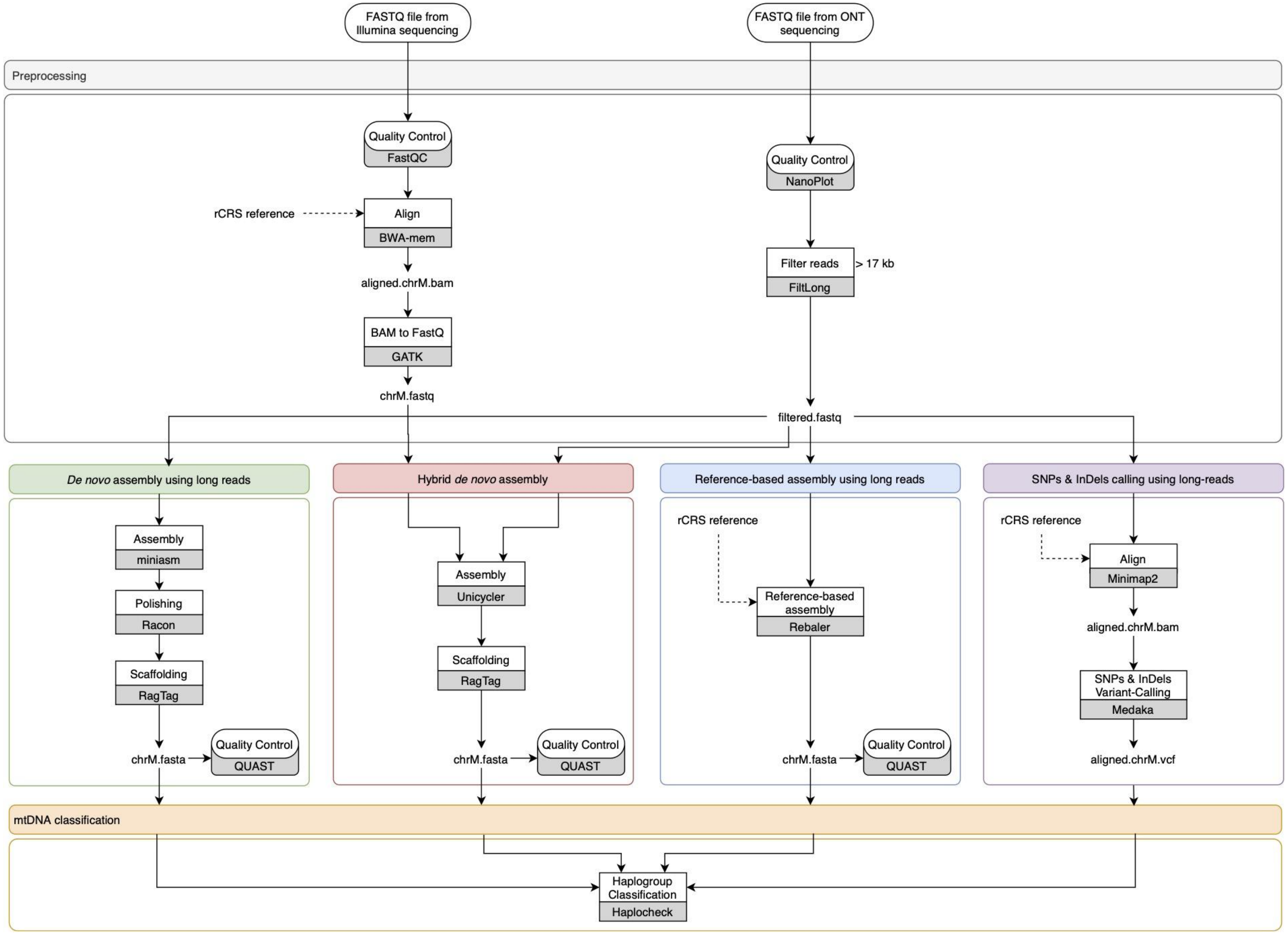

Table S1. Short-read sequencing summaries of the samples.

Samples	*Whole-Genome Sequencing*							*Whole-Exome Sequencing*						
	Protocol	*Mapped reads*	*Duplicate reads*	*Mapping Quality*	*Base Quality*	*Depth*	*Variants*	*Protocol*	*Mapped reads*	*Duplicate reads*	*Mapping Quality*	*Base Quality*	*Depth*	*Variants*
CAN01	Nextera-DNA-Prep	269,488	77,240	59.98	26.00	1227	42	Nextera-DNA-Exome	15,273	2,438	59.96	29.10	58	42
CAN02	Nextera-DNA-Prep	239,511	65,285	59.99	29.20	1466	11	Nextera-DNA-Exome	13,499	1,809	59.97	29.20	53	10
CAN03	Nextera-DNA-Prep	225,038	63,872	59.96	28.90	1293	14	Nextera-DNA-Exome	3,299	950	60.00	28.70	18	6
CAN04	Nextera-DNA-Prep	239,742	56,631	59.95	27.90	1589	10	Nextera-DNA-Exome	16,786	2,296	59.98	29.30	66	9
CAN05	Nextera-DNA-Prep	140,980	27,795	59.98	28.80	974	28	Nextera-DNA-Exome	9,925	1,274	59.98	29.30	39	25
CAN06	Nextera-DNA-Prep	209,422	86,736	59.98	28.60	977	44	Nextera-DNA-Exome	15,609	2,394	59.98	29.40	60	44
CAN07	Nextera-DNA-Prep	202,279	53,246	59.98	29.20	676	14	Nextera-DNA-Exome	17,727	3,247	59.98	29.20	66	14
CAN08	Nextera-DNA-Prep	102,834	22,765	59.97	28.70	661	41	Nextera-DNA-Exome	2,456	702	60.00	29.20	14	8
CAN09	Nextera-DNA-Prep	288,083	92,888	59.99	29.40	880	37	Nextera-DNA-Exome	36,117	15,507	59.99	29.50	93	34
CAN10	Nextera-DNA-Prep	107,424	17,424	59.98	28.80	760	26	Nextera-DNA-Exome	6,501	1,082	59.97	28.70	25	26
CAN11	Nextera-DNA-Prep	486,352	184,988	59.97	28.60	2577	25	Nextera-DNA-Exome	5,452	1,783	60.00	29.10	29	17
CAN12	Illumina-DNA-Prep	96,563	15,042	59.95	28.10	724	25	Nextera-DNA-Exome	5,297	850	59.95	28.70	20	22
CAN13	Nextera-DNA-Prep	298,707	84,386	59.98	29.00	1822	25	Nextera-DNA-Exome	23,503	3,853	59.99	29.30	89	25
CAN14	Nextera-DNA-Prep	160,800	31,404	59.98	28.60	1130	12	Nextera-DNA-Exome	8,746	3,222	60.00	29.40	25	12
CAN15	Illumina-DNA-Prep	86,288	12,342	59.94	27.90	656	34	Nextera-DNA-Exome	6,208	1,168	59.98	29.50	23	31
CAN16	Nextera-DNA-Prep	145,985	52,881	59.95	27.00	786	33	Nextera-DNA-Exome	5,211	1,964	60.00	29.10	23	20
CAN17	Nextera-DNA-Prep	287,742	80,049	59.96	28.40	1691	12	Nextera-DNA-Exome	5,532	1,705	60.00	28.80	30	9
CAN18	Nextera-DNA-Prep	186,339	78,863	59.99	28.90	874	34	Nextera-DNA-Exome	5,056	2,026	60.00	29.30	22	11
CAN19	Nextera-DNA-Prep	158,760	34,517	59.97	28.60	1021	13	Nextera-DNA-Exome	2,345	698	59.98	28.10	13	4
CAN20	Nextera-DNA-Prep	198,085	48,750	59.99	29.20	1284	13	Nextera-DNA-Exome	9,765	1,655	59.98	36.80	37	12
CAN21	Nextera-DNA-Prep	183,878	37,268	59.96	28.30	1282	17	Nextera-DNA-Exome	6,736	965	59.99	36.60	26	17
CAN22	Nextera-DNA-Prep	466,404	196,793	59.98	29.10	1823	7	Nextera-DNA-Exome	3,911	1,212	59.99	29.20	22	1
CAN23	Nextera-DNA-Prep	149,371	32,434	59.99	28.60	1002	35	Nextera-DNA-Exome	9,810	1,422	59.99	29.20	38	35
CAN24	Nextera-DNA-Prep	75,746	10,746	59.97	28.70	554	34	Nextera-DNA-Exome	6,809	1,010	59.95	29.20	26	32
CAN25	Nextera-DNA-Prep	130,389	23,037	59.97	28.40	935	39	Nextera-DNA-Exome	6,707	1,184	59.99	29.30	25	37
CAN26	Illumina-DNA-Prep	145,401	30,564	59.96	28.40	1024	35	Nextera-DNA-Exome	10,105	1,900	59.99	29.50	37	32
CAN27	Illumina-DNA-Prep	106,440	16,750	59.94	28.00	796	39	Nextera-DNA-Exome	6,087	996	60.00	29.40	23	38
CAN28	Nextera-DNA-Prep	341,321	102,609	59.97	25.80	1594	10	Nextera-DNA-Exome	13,280	2,376	59.98	29.20	50	9
CAN29	Nextera-DNA-Prep	351,160	108,741	59.97	25.80	1618	15	Nextera-DNA-Exome	17,020	5,915	59.99	29.60	50	15
CAN30	Nextera-DNA-Prep	189,623	78,268	59.97	27.90	973	38	Nextera-DNA-Exome	4,347	1,745	60.00	29.20	19	11
CAN31	Nextera-DNA-Prep	143,579	32,325	59.98	29.20	950	26	Nextera-DNA-Exome	10,019	2,159	59.93	28.70	36	25
CAN32	Nextera-DNA-Prep	116,947	21,154	59.98	28.60	824	26	Nextera-DNA-Exome	7,947	1,362	59.98	29.10	30	25
CAN33	Nextera-DNA-Prep	138,396	38,670	59.99	29.30	649	31	Nextera-DNA-Exome	4,038	1,204	60.00	29.10	23	17
CAN34	Nextera-DNA-Prep	130,484	23,846	59.98	28.80	913	43	Nextera-DNA-Exome	14,569	2,380	59.98	29.00	55	42
CAN35	Illumina-DNA-Prep	119,570	22,308	59.96	28.50	867	40	Nextera-DNA-Exome	5,709	732	59.97	35.10	23	38
CAN36	Illumina-DNA-Prep	198,697	40,465	59.95	28.30	1402	26	Nextera-DNA-Exome	15,194	2,908	59.99	29.50	56	26

Table S2. Multicollinearity test results (Pearson correlation coefficients) based on cross-correlation among the selected parameters estimated from the BAM files for short-read A) WGS and B) WES datasets. The statistical significance is indicated by asterisks: * *p* ≤ 0.05 | ** *p* ≤ 0.01 | *** *p* ≤ 0.001

a)

R square WGS	***Mapped reads***	***Duplicate reads***	***Mapping Quality***	***Base quality***	***Coverage***
Mapped reads	1.000	0.959***	0.190	-0.197	0.877***
Duplicates reads	0.959***	1.000	0.224	-0.135	0.772***
Mapping quality	0.190	0.224	1.000	0.382*	0.018
Base quality	-0.197	-0.135	0.382*	1.000	-0.172
Depth	0.877***	0.772***	0.018	-0.172	1.000

b)

R square WES	***Mapped reads***	***Duplicate reads***	***Mapping Quality***	***Base quality***	***Coverage***
Mapped reads	1.000	0.837***	-0.107	-0.040	0.960***
Duplicates reads	0.837***	1.000	0.095	-0.076	0.677***
Mapping quality	-0.107	0.095	1.000	-0.006	-0.113
Base quality	-0.040	-0.076	-0.006	1.000	-0.064
Depth	0.960***	0.677***	-0.113	-0.064	1.000

Table S3. Mitochondrial DNA haplogroup consensus and concordance rate among the 11 tools based on short-read WGS for the samples assessed.

Samples	*Haplogroup*	*Concordance rate*
CAN01	J2a2d	100.00
CAN02	H7	100.00
CAN03	H1ao1	100.00
CAN04	H1t	92.31
CAN05	U6b1a	100.00
CAN06	J2a2d	92.31
CAN07	H6a1b2	100.00
CAN08	L3d1b3a	92.31
CAN09	U4c1	69.23
CAN10	U6b1a	92.31
CAN11	U6b1a	92.31
CAN12	U6b1a	100.00
CAN13	U6b1a	100.00
CAN14	H1e1a	92.31
CAN15	U4c1	69.23
CAN16	T1a	92.31
CAN17	H1+16189	69.23
CAN18	K1a4a1	100.00
CAN19	H1e1a	100.00
CAN20	H1au	92.31
CAN21	HV+16311	69.23
CAN22	H	69.23
CAN23	K1a1b1	92.31
CAN24	K1a14	92.31
CAN25	J1c2c1	92.31
CAN26	J1c2	92.31
CAN27	T2c1d+152	69.23
CAN28	H1cf	69.23
CAN29	H6a1a	92.31
CAN30	U5b2b3a	92.31
CAN31	U6b1a1	100.00
CAN32	U6b1a	92.31
CAN33	X3a	92.31
CAN34	L3f1b1a	92.31
CAN35	L3b1a+@16124	69.23
CAN36	U6b1a1	100.00

Table S4. Per sample mtDNA haplogroup classifications for the 11 tools based on short-read A) WGS and B) WES datasets.

a)

Samples	***Haplogrep VCF***	***Haplogrep FASTA***	***Haplocheck BAM***	***Haplocheck VCF***	***Haplotracker FASTA***	***HaploGrouper VCF***	***Phy-Mer FASTA***	***Phy-Mer BAM***	***EMMA FASTA***	***MitoSuite BAM***	***Haplofind FASTA***	***MixEmt BAM***	***MitoTool FASTA***	***James Lick's FASTA***
CAN01	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2a	J2a2d	J2a2d
CAN02	H7	H7	H7	H7	H7	H7	H7	H7	H7	H27e	H7	H22	H7	H7
CAN03	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1	H1ao1
CAN04	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t2	H1t	H1t
CAN05	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN06	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2a	J2a2d	J2a2d
CAN07	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2b	H6a1b2	H6a1b2
CAN08	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d1b3a	L3d4a	L3d1b3a	L3d1b3a
CAN09	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1a	U4c1a	U4c1	U4c1	U4c1	U4a2g	U4c1a	U4c1
CAN10	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a1	U6b1a	U6b1a
CAN11	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a1	U6b1a	U6b1a
CAN12	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN13	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN14	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1a4	H1e1a	H1e1a
CAN15	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1a	U4c1a	U4c1	U4c1	U4c1	U4c1a	U4c1a	U4c1
CAN16	T1a	T1a	T1a	T1a	T1a	T1a	T1a	T1a	T1a	T1a	T1a	T1a1b1	T1a	T1a
CAN17	H1+16189	H1+16189	H1+16189	H1+16189	H1+16189	H1+16189	H1-16189	H1-16189	H1+16189	B	H1	B4a1a1ab	H1	H1+16189
CAN18	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1	K1a4a1
CAN19	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a1	H1e1a	H1e1a
CAN20	H1au	H1au	H1au	H1au	H1au	H1au	H1au	H1au	H1au	H1au	H1au	H28	H1au	H1au
CAN21	HV+16311	HV+16311	H+16311	HV+16311	HV+16311	HV+16311	HV-16311	HV-16311	HV+16311	B	HV	R0a'b	HV	HV+16311
CAN22	H	H	H	H	H	H77	H	H-16291	H	H27e	H	H3b	H	H
CAN23	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1b	K1a1b1	K1a1b1
CAN24	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a5a	K1a14	K1a14
CAN25	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2h	J1c2c1	J1c2c1
CAN26	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c4c	J1c2	J1c2
CAN27	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d-152	T2c1d-152	T2c1d+152	T2c1d2	T2c1d2	T2c1d[1]	T2c1d	T2c1d+152
CAN28	H1cf	H1cf	H1cf	H1cf	H1cf	H1cf	H1	H1	H1cf	H1cf	H1cf	H3ac	H1	H1cf
CAN29	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a2b1	H6a1a	H6a1a
CAN30	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b3a	U5b2b	U5b2b3a	U5b2b3a
CAN31	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a	U6b1a1	U6b1a1
CAN32	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a1	U6b1a	U6b1a
CAN33	X3a	X3a	X3a	X3a	X3a	X3a	X3a	X3a	X3a	X3a	X3a	U4b1a1a1	X3a	X3a
CAN34	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a
CAN35	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a-@16124	L3b1a-@16124	L3b1a+@16124	L3b1a11	L3b1a	L3b1a3	L3b1a	L3b1a-@16124
CAN36	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1

b)

Samples	***Haplogrep VCF***	***Haplogrep FASTA***	***Haplocheck BAM***	***Haplocheck VCF***	***Haplotracker FASTA***	***HaploGrouper VCF***	***Phy-Mer FASTA***	***Phy-Mer BAM***	***EMMA FASTA***	***MitoSuite BAM***	***Haplofind FASTA***	***MixEmt BAM***	***MitoTool FASTA***	***James Lick's FASTA***
CAN01	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d
CAN02	H7	H7	H7	H7	H7	H7	H7	H7	H7	H27e	H7	H7	H7	H7
CAN03	H1ao	H1ao	H1ao1	H1ao	H2a2a1	H2a2a1	H2a2a1	H1x	H2a2a1	H1ao1	H2a2a1	H1ao1	H2a2a1	H2a2a1
CAN04	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t	H1t
CAN05	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN06	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d	J2a2d
CAN07	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2	H6a1b2
CAN08	L3d	L3d	L3d1b3a	L3d	H2a2a1	H2a2a1	H2a2a1	L3d1b1a	H2a2a1	L3d1b3a	H2a2a1	L3d1b3a	H2a2a1	H2a2a1
CAN09	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1a	U4c1a	U4c1	U4c1	U4c1	U4c1	U4c1a	U4c1
CAN10	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN11	U6b1a	U6b1a	U6b1a	U6b1a	U6+16311	U6a'b'd T16311C	U6a'b'd-16311	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6a'b'd	U6a'b'd
CAN12	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN13	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN14	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a	H1e1a
CAN15	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1	U4c1a	U4a2c	U4c1	U4c1	U4c1	U4c1	U4c1a	U4c1
CAN16	T1a	T1a	T1a	T1a	T1a	T1a	T1a	T1a6	T1a	T1a	T1a	T1a	T1a	T1
CAN17	H2	H2	H1+16189	H2	H1a	H1a	H1a	H1a2	H2	B	H1a	H1	H1	H2
CAN18	K1a4	K1a4	K1a4a1	K1a4	H2a2	H2a2	H2a2a	K1a4a1	H2a2a	K1a4a1	H2a2a	K1a4a1	H2a2	H2a2a
CAN19	H2a	H2a	H1e1a	H2a	H2a	H2a	H2a	H1e1b1a	H2a	H1e1a	H2a	H1e1a	H2a	H2a
CAN20	H1au	H1au	H1au	H1au	H1au	H1au	H1au	H1-16311	H1au	H1au	H1au	H1au	H1au	H1au
CAN21	HV+16311	HV+16311	H+16311	HV+16311	HV+16311	HV+16311	HV-16311	HV-16311	HV+16311	B	HV	HV	HV	HV+16311
CAN22	H2	H2	H	H2	H2a2a1	H2a2a1	H2a2a1	H30a	H2a2a1	H27e	H2a2a1	H	H2a2a1	H2a2a1
CAN23	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	k1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1	K1a1b1
CAN24	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14	K1a12	K1a14	K1a14	K1a14	K1a14	K1a14	K1a14
CAN25	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1	J1c2c1
CAN26	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2	J1c2
CAN27	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d+152	T2c1d-152	T2c1d-152	T2c1d+152	T2c1d2	T2c1d2	T2c1d2	T2c1d	T2c1d+152
CAN28	H1cf	H1cf	H1cf	H1cf	H1cf	H1cf	H1	H1	H1cf	H1cf	H1cf	H1cf	H1	H1cf
CAN29	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a	H6a1a
CAN30	U5b2b3	U5b2b3	U5b2b3a	U5b2b3	H2a2a1	H2a2a1	H2a2a1	U5b2b3a	H2a2a1	U5b2b3a	H2a2a1	U5b2b3a	H2a2a1	H2a2a1
CAN31	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1
CAN32	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a	U6b1a
CAN33	X	X	X3a	X	X	X	X	X3a	X	X3a	X	X3a	X	X
CAN34	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a	L3f1b1a
CAN35	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a+@16124	L3b1a-@16124	L3b1a-@16124	L3b1a+@16124	L3b1a11	L3b1a	L3b1a	L3b1a	L3b1a+@16124
CAN36	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1	U6b1a1

Table S5. BGLMM model results for WGS and WES datasets. Summary of values obtained with the haplogroup classification tool as fixed factor. Mapped reads, mapping quality, and base quality were introduced as covariates. Discordance was used as the response variable and the sample was used as a random factor. The statistical significance is marked with asterisks: * $p \leq 0.05$ | ** $p \leq 0.01$ | *** $p \leq 0.001$

Application	*Tool*	*Estimate*	*Std.Error*	*zvalue*	*Pr(>|z|)*	
WGS	Haplotracker FASTA	-3.16	0.66	-4.81	0.00	***
-	Phy-Mer FASTA	0.31	0.59	0.52	0.60	
-	EMMA FASTA	-2.20	2.41	-0.91	0.36	
-	Haplofind FASTA	0.56	0.57	0.98	0.33	
-	MitoTool FASTA	1.13	0.55	2.05	0.04	*
-	Haplogrep FASTA	-2.18	2.35	-0.93	0.35	
-	JamesLick's FASTA	-2.16	2.32	-0.93	0.35	
-	HaploCheck BAM	-2.20	2.39	-0.92	0.36	
-	MixEmt BAM	3.55	0.66	5.35	0.00	***
-	MitoSuite BAM	0.76	0.56	1.36	0.17	
-	Phy.mer BAM	0.55	0.57	0.96	0.34	
-	HaploCheck VCF	-2.14	2.27	-0.94	0.35	
-	Haplogrep VCF	-2.13	2.25	-0.95	0.34	
-	HaploGrouper VCF	-0.43	0.71	-0.61	0.54	
-	Mapped Reads	0.37	0.30	1.23	0.22	
-	Mapping Quality	0.06	0.32	-1.86	0.06	.
-	Base Quality	-0.09	0.51	0.17	0.87	
WES	Haplotracker FASTA	-6.84	2.82	-2.42	0.02	*
-	Phy-Mer FASTA	0.47	0.48	0.98	0.33	
-	EMMA FASTA	-0.36	0.51	-0.70	0.48	
-	Haplofind FASTA	0.22	0.49	0.45	0.66	
-	MitoTool FASTA	0.97	0.49	1.98	0.05	*
-	Haplogrep FASTA	-0.55	0.52	-1.07	0.29	
-	JamesLick's FASTA	0.14	0.49	0.28	0.78	
-	HaploCheck BAM	-4.38	1.99	-2.20	0.03	*
-	MixEmt BAM	-1.13	0.50	-2.25	0.02	*
-	MitoSuite BAM	-0.63	0.48	-1.32	0.19	
-	Phy.mer BAM	0.27	0.45	0.60	0.55	
-	HaploCheck VCF	-0.59	0.52	-1.13	0.26	
-	Haplogrep VCF	-0.57	0.52	-1.09	0.27	
-	HaploGoruper VCF	-0.13	0.50	-0.26	0.80	
-	Mapped Reads	-5.96	3.55	-1.68	0.09	.
-	Mapping Quality	1.16	0.39	2.97	0.00	**
-	Base Quality	0.15	0.24	0.61	0.54	