

Supplementary Material For:

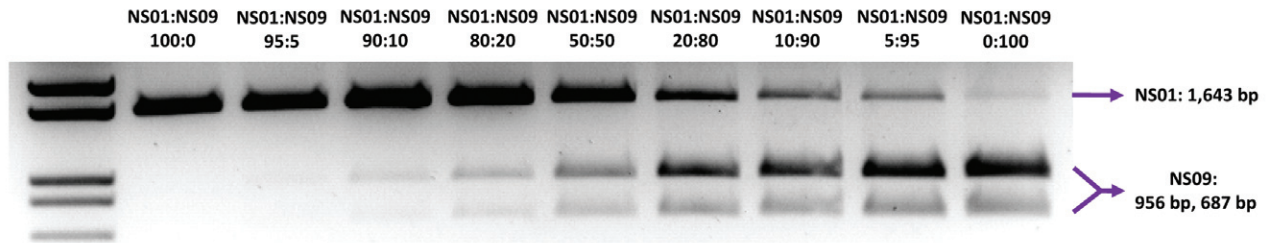
Characterization of mitochondrial DNA heteroplasmy using a parallel sequencing system

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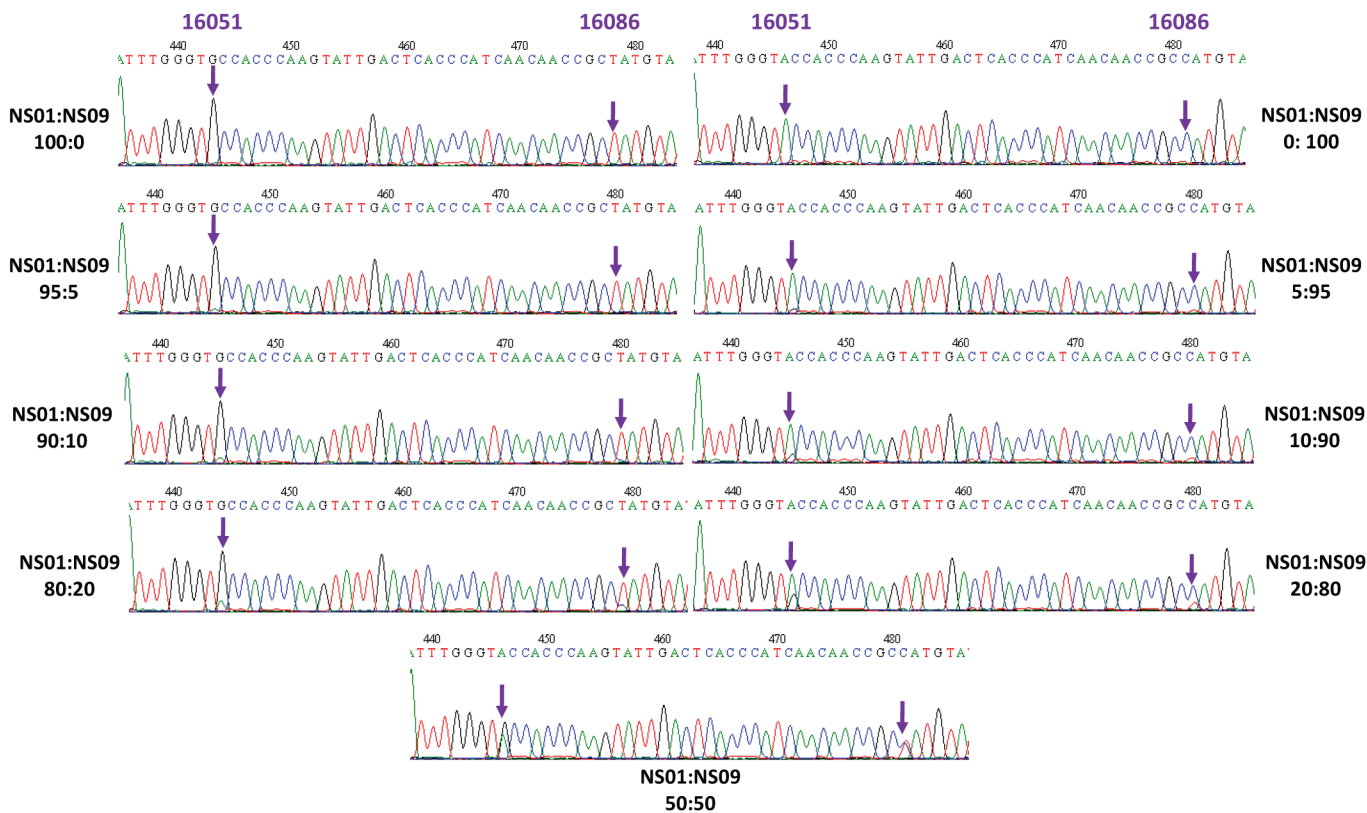
Supplementary Figure S1. Comparison of parallel sequencing to RFLP.

Supplementary Table S1. Detected heteroplasmy levels from the mixture tests for the 56 expected variant positions between NS01 and NS09

rCRS position	rCRS base	NS01 base	NS09 base	Test I						Test II				
				1% raw	1% final	5% final	10% final	20% final	50% final	1% final	5% final	10% final	20% final	50% final
143	G	G	A	1.1	ND	5.6	7.5	17.3	47.6	ND	4.8	8.7	19.2	49.3
146	T	T	C	ND	ND	5.5	8.2	18.4	49	ND	4.6	8.4	20.1	50.6
150	C	T	C	2.0	ND	5.1	9.5	21.7	53.2	ND	4.3	8.9	19.4	47.8
152	T	T	C	1.5	ND	4.0	9.1	19.9	49.3	ND	4.4	9.4	20.0	49.2
195	T	T	C	1.2	ND	4.4	9.3	19.8	50.7	ND	3.3	10.5	18.5	51.0
493	A	G	A	1.3	ND	5.4	11.3	18.9	48.1	ND	6.5	10.6	22.8	53.4
721	T	C	T	1.5	0.5	4.5	11.5	20.7	49.5	ND	4.5	10.4	23.8	47.5
750	A	A	G	1.0	ND	5.8	11.3	22.1	49.8	1.0	5.6	10.9	21.8	46.9
769	G	G	A	1.2	ND	3.9	11.2	22.0	49.6	ND	4.3	10.8	20.3	49.1
1018	G	G	A	1.5	ND	7.8	12.2	25.2	54.3	ND	5.4	10.2	17.8	45
2352	T	C	T	0.8	ND	5.7	11.3	19.4	50.2	0.9	4.3	8.2	18.6	47.3
2416	T	T	C	1.0	ND	4.5	10.6	21.6	48.7	0.6	5.1	10.5	20.2	47.2
2755	A	A	G	1.3	ND	5.4	11.6	22.4	52.9	1.0	4.5	9.4	19.6	48.4
2789	C	C	T	1.2	ND	5.7	13.0	20.7	51.2	ND	5.8	12.3	20.3	50.2
3010	G	G	A	1.4	ND	4.7	9.7	19.5	45.2	1.0	6.7	9.7	21.5	51.0
3594	C	C	T	1.3	ND	5.9	10.7	20.8	50.5	ND	4.3	11.1	23.3	51.2
3915	G	A	G	1.2	ND	5.7	8.9	18.7	48.1	1.7	6.1	9.3	19.2	45.9
4104	A	A	G	1.4	ND	4.6	10.2	19.4	48.6	ND	5.2	11.7	21.4	46
5262	G	A	G	1.5	1.5	4.6	11.0	20.7	51.1	1.4	5.6	8.3	19.3	48.3
5584	A	G	A	1.3	ND	5	9.4	21.7	47.9	0.8	5.0	10.9	22.4	51.1
6663	A	A	G	1.0	ND	5.5	9.9	20.5	50.0	ND	5.3	11.4	20.2	49.9
7175	T	T	C	1.4	1.2	6.1	10.1	22.1	50.3	ND	5.0	10.5	20.8	48.3
7256	C	C	T	1.1	ND	5.4	11.2	19.9	49.5	ND	3.1	9.1	19.6	48.1
7274	C	C	T	1.2	ND	4.0	9.9	20.2	48.1	ND	5.0	10.4	21.6	50.8
7521	G	G	A	1.2	ND	4.1	9.5	20.2	48.1	ND	3.5	10.0	17.4	50.6
7771	A	A	G	1.2	ND	5.1	9.3	21.7	50.3	0.9	4.1	9.0	19.5	47
8206	G	G	A	1.4	ND	4.0	9.5	20.4	47.4	ND	4.8	8.7	19.7	50.2
9221	A	A	G	1.4	ND	5.5	11.3	20.1	50.5	ND	5.1	10.8	18.6	47.1
9401	C	C	T	1.2	ND	3.7	9.6	22.2	50.9	0.6	5.3	10.6	20.2	46.4
9596	A	A	G	ND	ND	1.8	3.4	6.3	14.4	ND	1.6	3.4	6.5	54
9656	T	T	C	1.2	ND	4.9	13.2	24.1	53.5	ND	6.0	10.5	19.7	47.2
10115	T	T	C	ND	ND	4.7	10.8	20	49.9	ND	2.0	12.3	22.9	47.2
10819	A	G	A	1.2	0.8	5.9	10.1	19.3	48.3	0.8	2.1	10.6	16.5	45.4
11257	C	T	C	1.2	0.8	4.8	9.9	20.2	50	0.8	5.2	10.5	19.6	48.1
11914	G	G	A	1.3	ND	5.2	9.5	19.0	45.2	1.3	5.5	9.9	21.0	48.4
11944	T	T	C	1.3	ND	4.0	8.5	18.0	44.9	ND	5.8	9.7	22.5	50.9
12693	A	A	G	1.6	1.6	5.2	9.5	21.7	49.4	1.0	3.1	9.5	20.3	48.1
13590	G	G	A	ND	ND	4.9	9.6	19.6	48.8	0.8	4.8	10.1	19.9	48.6
13650	C	C	T	1.0	ND	4.7	11	20.4	49.3	ND	3.8	8.2	19.5	47.4
13749	C	T	C	1.4	ND	5.7	9.9	21.1	49.1	0.7	2.8	9.1	19.6	46.2
13803	A	A	G	1.2	ND	4.9	10.4	18.6	50.1	ND	5.5	8.8	17.7	49.2

Supplementary Table S1 (continued). Detected heteroplasmy levels from the mixture tests for the 56 expected variant positions between NS01 and NS09

rCRS position	rCRS base	NS01 base	NS09 base	Test I								Test II			
				1% raw	1% final	5% final	10% final	20% final	50% final	1% final	5% final	10% final	20% final	50% final	
14212	T	C	T	1.2	ND	6.1	10.1	18.2	46.2	1.1	4.9	9.4	19.8	46.7	
14566	A	A	G	1.6	ND	6.3	10.8	22.1	50.4	0.9	5.1	10.8	19.1	46.9	
15262	T	C	T	1.4	1.0	4.9	9.1	22.0	46.9	ND	3.5	13.0	22.3	55.6	
15784	T	T	C	1.2	1.0	5.5	9.5	20.6	46.3	1.0	3.5	9.4	21.3	47.6	
15940	T	T	C	1.7	ND	4.4	10.3	19.8	49.6	ND	4.0	11.2	23.2	50.7	
16051	A	G	A	1.6	ND	4.6	10.2	20.1	47.9	ND	3.9	10.8	23.2	51.8	
16086	T	T	C	1.1	ND	4.2	8.8	16.8	30.8	ND	3.2	6.4	13.5	34.6	
16264	C	T	C	1.5	1.3	3.6	10.4	20.3	48.2	ND	5.7	11.0	19.9	49.5	
16278	C	C	T	1.2	ND	4.8	11.2	18.5	46.7	0.6	6.2	9.5	19.6	45.8	
16294	C	C	T	1.0	ND	5.2	10.6	18.5	46.8	ND	5.1	9.4	19.3	46.4	
16309	A	A	G	ND	ND	6.4	10.4	20.4	48.4	ND	4.6	9.9	19.8	48.0	
16390	G	G	A	1.4	ND	2.7	8.4	17.8	44.9	ND	5.6	9.4	20.0	47.9	
16519	T	C	T	1.6	1.2	6.4	10.4	18.6	47.3	1.1	3.8	8.8	21.3	48.3	
Mean				1.30	1.09	4.98	10.07	20.00	48.03	0.95	4.60	9.86	19.91	48.43	
sd				0.22	0.34	0.97	1.44	2.49	5.61	0.27	1.13	1.48	2.60	2.92	
523	A	del	A	29.2	ND	3.4	10.9	16.4	56.6	ND	2.8	7.0	14.2	29.1	
524	C	del	C	37.2	ND	3.5	11.7	15.8	54.5	ND	2.8	6.8	14.1	27.5	
ND, not detected.															



Supplementary Figure S2. Comparison of parallel sequencing to direct sequencing.

Supplementary Table S2. Comparison of parallel sequencing to RFLP

rCRS position	rCRS base	NS01 base	NS09 base	Test 1					Test 2				
				1%	5%	10%	20%	50%	1%	5%	10%	20%	50%
16278	C	C	T	ND	4.8	11.2	18.5	46.7	0.6	6.2	9.5	19.6	45.8

ND, not detected.

Supplementary Table S3. Comparison of parallel sequencing to direct sequencing

rCRS position	rCRS base	NS01 base	NS09 base	Test 1					Test 2				
				1%	5%	10%	20%	50%	1%	5%	10%	20%	50%
16051	A	G	A	ND	4.6	10.2	20.1	47.9	ND	3.9	10.8	23.2	51.8
16086	T	T	C	ND	4.2	8.8	16.8	30.8	ND	3.2	6.4	13.5	34.6

ND, not detected.