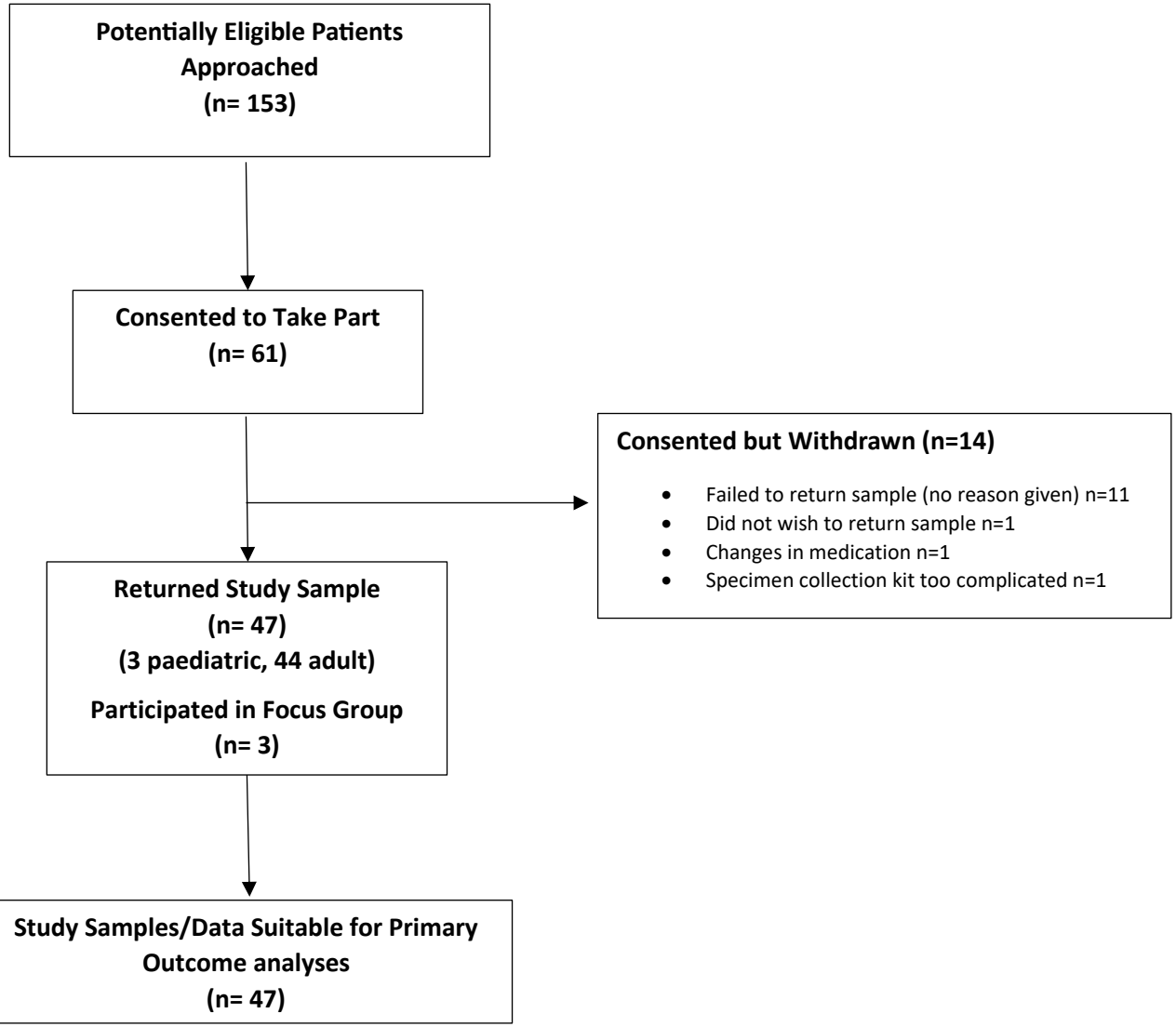
FiND ISRCTN Results Upload

Participant Flow

Identification/Invitation



Baseline Characteristics Table

	Adult (N=44)	**Paediatric (N=3)**
Age (years)	52.2	7.67
Sex	Male (N=13 (29.5%)), Female (N=31 (70.5%))	Male (N=2 (66.6%)), Female (N=1 (33.3%))
Asymptomatic	3 (6.8%)	2 (66.6%)
Diabetes	24 (54.5%)	0 (0%)
GI/renal impairments	25 (56.8)	1 (33.3%)
Deafness/ hearing impairments	33 (75%)	0 (0%)
Muscular impairments/ exercise intolerance	12 (27.3%)	0 (0%)
Fatigue	11 (25%)	0 (0%)
Ataxia	9 (20.5%)	0 (0%)
Visual impairments	9 (20.5%)	0 (0%)
Cardiac impairments	7 (15.9%)	0 (0%)

Outcome Measures Table

Objectives	
Primary	• Validate the use of faecal specimens as a novel diagnostic concept for mtDNA-related mitochondrial disease against historic samples of muscle, blood, urine or buccal derived DNA using pyrosequencing.
Secondary	• Validate the use of Whole Genome Sequencing of mtDNA extracted from faecal samples to detect mtDNA variants that may be present. • Collect patient feedback to assess current conceptions and feelings towards current diagnostic approaches and the potential use of faecal samples for diagnostic purposes.

ID	Historic muscle heteroplasmy (%)	Historic blood heteroplasmy (%)	Age adjusted blood (%)	Historic urine heteroplasmy (%)	Urine adjusted (%)	Historic buccal heteroplasmy (%)	stool heteroplasmy (%)	WGS heteroplasmy (%)
A001	11	3	15	14	16	2	25	
A002	n/a	20	62	56	71	n/a	64	
A004	n/a	37	84	69	83	54	67	
A005	n/a	33	94	85	94	59	80	
A006	70	21	100	41	54	n/a	77	
A007	n/a	40	100	23	29	14	52	
A008	n/a	5	24	25	31	n/a	41	
A009	n/a	29	81	47	61	38	70	
A010	67	23	100	72	64	n/a	74	81.5
A011	n/a	30	100	60	75	n/a	75	
A012	n/a	6	42	41	54	n/a	68	
A015	n/a	27	98	85	83	57	74	
A016	n/a	23	100	73	87	n/a	67	
A017	n/a	22	69	n/a	n/a	n/a	63	
A018	n/a	24	83	86	84	n/a	76	
A020	86	55	100	76	89	64	81	
A021	n/a	20	65	67	57	n/a	64	
A024	n/a	28	100	53	68	n/a	78	
A025	n/a	14	66	66	55	n/a	78	

A026	87	22	89	84	81	n/a	80	
A028	80	24	100	84	81	n/a	73	
A029	80	22	98	85	83	n/a	81	86
A033	n/a	13	38	n/a	n/a	n/a	51	
A034	n/a	6	36	51	66	n/a	61	
A035	n/a	16	44	36	47	n/a	62	
A036	67	n/a	n/a	66	55	n/a	42	
A037	74	4	16	12	13	n/a	29	
A038	n/a	n/a	n/a	20	24	n/a	30	
A040	68	34	100	82	78	36	75	
A041	n/a	33	82	46	60	n/a	68	
A042	n/a	9	36	41	54	n/a	45	
A043	n/a	17	89	74	87	n/a	64	
A044	n/a	4	27	21	26	n/a	57	
A045	n/a	36	82	80	91	42	70	
A046	n/a	24	100	77	71	n/a	79	
A047	n/a	26	85	48	62	n/a	73	
A048	n/a	28	85	72	86	n/a	73	
A049	n/a	14	69	n/a	n/a	n/a	76	
A050	n/a	9	28	28	36	n/a	32	
A051	71	25	79	76	89	n/a	73	62.2
A053	n/a	16	58	55	70	n/a	61	
A054	76	19	82	47	61	n/a	79	
A055	n/a	8	46	48	62	n/a	63	
A056	84	23	100	74	87	n/a	71	
P001	n/a	75	n/a	n/a	n/a	n/a	82	
P004	51	52	n/a	57	72	n/a	70	74.2
P005	n/a	79	n/a	n/a	n/a	n/a	81	

Adverse Events Table

There were no adverse events associated with this study.