Title: The Palestinian primary ciliary dyskinesia population: first results of the diagnostic, and genetic spectrum

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Supplementary table and Figures

Supplementary table 1. Gene variants, familial genetics and screening method for 45 individuals (from 40 families) positively genetically diagnosed in the study. Panel, targeted gene panel; WES, whole exome sequencing (Agilent SureSelect Human All Exon V6); MTD, microtubular disorganisation; IDA, inner dynein arm defect; ODA, outer dynein arm defect; CC, central pair complex defect; T, transposition of peripheral microtubules. [§]A total 51 individuals carried causal variants, with 6 individuals listed at the bottom carrying homozygous VUS (variant of unknown significance) not regarded as diagnostic without further evidence.

Supplementary Figure 1 Outcomes of test results for people with a clinical suspicion of PCD. The order of tests was usually as suggested by the flow diagram, but on occasions the order was different e.g. nNO sometimes followed TEM. ⊕= PCD confirmed according to ERS guidelines.

Supplementary Figure 2: Genetic diagnoses associated with different PCD disease features in Palestine PCD patients

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Family Study ID	Gene	Variant (all homozygous)[§]	No. affected children with homozygous mutations	Parents available and consistent for segregation	Consistent segregation in # additional siblings	Sequencing method	TEM consistent
0-1	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	4	Panel	MTD+IDA
0-2	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	5	Panel	MTD
0-8	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	1	Panel	MTD
0-9	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	1	Panel	MTD+IDA
0-11	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	5	Panel	MTD+IDA
0-20	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	0	Panel	MTD+IDA
0-21	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	2	Panel	MTD+IDA
0-25	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	1	Allele-specific PCR	MTD+IDA
0-31	*CCDC39*	c.1871_1872del; p.(Ile624Lysfs*3)	1	Mat and Pat	0	Allele-specific PCR	MTD+IDA
0-35	*CCDC39*	c.2190del; p.(Glu731Asnfs*31)	1	Mat and Pat	2	WES	MTD+IDA

0-6	*CCDC40*	c.48A>G; p.(Gly16Gly)	4	Mat and Pat	2	Panel	MTD+IDA
0-36	*CCNO*	c.381+5G>C	1	Mat and Pat	1	WES	Oligocilia
0-39	*CCNO*	c.381+5G>C	1	Mat and Pat	5	WES	Oligocilia
0-16	*DNAAF4 (DYX1C1)*	c.384_390del; p.(Tyr128*)	1	Mat only	0	Panel	ODA+IDA
0-30	*DNAAF4 (DYX1C1)*	c.384_390del; p.(Tyr128*)	1	Mat and Pat	4	WES	ODA
0-40	*DNAAF4 (DYX1C1)*	c.384_390del; p.(Tyr128*)	1	Mat and Pat	3	WES	ODA+IDA
0-4	*DNAAF11 (LRRC6)*	c.436G>C; p.(Asp146His)	1	Mat and Pat	2	Panel	ODA+IDA
0-15	*DNAAF11 (LRRC6)*	c.436G>C; p.(Asp146His)	1	Mat only	4	Panel	ODA+IDA
0-18	*DNAAF11 (LRRC6)*	c.436G>C; p.(Asp146His)	1	Mat only	0	Panel	ODA+IDA
0-26	*DNAAF11 (LRRC6)*	c.436G>C; p.(Asp146His)	1	Mat and Pat	3	WES	ODA
0-42	*DNAAF11 (LRRC6)*	c.436G>C; p.(Asp146His)	1	Mat and Pat	0	Allele-specific PCR	ODA+IDA
0-23	*DNAH5*	c.10050G>A; p.(Trp3350*)	2	Mat and Pat	2	Panel	ODA
0-24	*DNAH11*	c.6727C>T; p.(Arg2243*)	1	Mat and Pat	3	Panel	Normal
0-7	*DNAH11*	c.12646G>T; p.(Glu4216*)	1	Mat and Pat	1	Panel	Normal
0-22	*DNAH11*	c.13240dup; p.(Thr4414Asnfs*34)	1	Mat and Pat	4	Panel	Normal
0-28	*DNAH11*	c.13436_13440dup; p.(Tyr4481Leufs*7)	1	Mat and Pat	5	WES	Normal

0-14	*DRC1*	c.1521_1524del; p.(Glu508Alafs*4)	1	Mat and Pat	3	WES	Normal
0-19	*DRC1*	c.1521_1524del; p.(Glu508Alafs*4)	1	Mat and Pat	2	WES	Normal
0-5	*ODAD3 (CCDC151)*	c.850C>T; p.(Gln284*)	1	Mat and Pat	2	Panel	ODA
0-29	*RSPH4A*	c.367del; p.(Pro123Leufs*44)	1	Mat and Pat	4	WES	CC+T
0-37	*RSPH4A*	c.72G>A; p.(Trp24*)	1	No parents	0	WES	CC+T
0-27	*RSPH9*	c.800-802del; p.(Lys268del)	1	Mat and Pat	2	WES	CC+T
0-32	*RSPH9*	c.800-802del; p.(Lys268del)	1	No parents	0	WES	CC
0-33	*RSPH9*	c.800-802del; p.(Lys268del)	1	Mat only	0	WES	CC+T
0-34	*SPAG1*	c.742C>T; p.(Arg248*)	1	Mat and Pat	4	WES	ODA
0-3	*CCDC103*	c.104G>C; p.(Arg35Pro) (VUS)	1	Mat and Pat	2	Panel	ODA+IDA
0-10	*DNAH11*	c.563T>C; p.Met188Thr (VUS)	1	Mat and Pat	0	Panel	Normal
0-12	*DNAH11*	c.563T>C; p.Met188Thr (VUS)	1	Mat only	0	Panel	Insufficient
0-17	*DNAL1*	c.285_287del; p.(Glu97del) (VUS)	1	Mat and Pat	5	WES	ODA
0-13	*RSPH9*	c.760del; p.(Arg254Alafs*76) (VUS)	1 (+1, as father also affected)	Mat and Pat (father also homozygous)	0	Panel	CC+T

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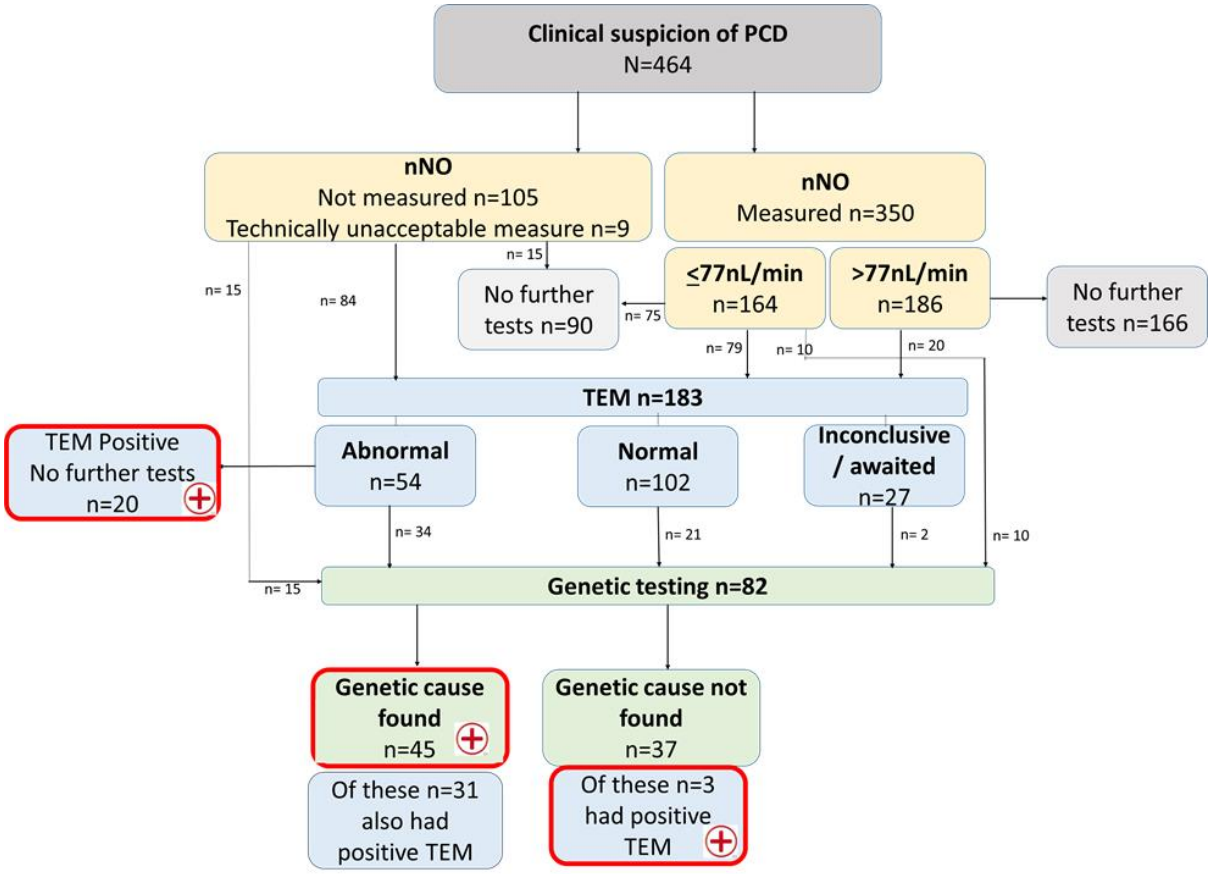

Supplementary Figure 2: Genetic diagnoses associated with different PCD disease features in Palestine PCD patients.