**Supplementary table 1: Genes included in the panel used.**

|  |  |  |  |
| --- | --- | --- | --- |
| **Gene** | **Ensembl ID** | **Genomics England primary ciliary disorders gene panel RAG rating** | **Literature** |
| *CCDC103* | ENSG00000167131 | Green | (1) |
| *CCDC39* | ENSG00000284862 | Green | (2) |
| *CCDC40* | ENSG00000141519 | Green | (3) |
| *CCDC65* | ENSG00000139537 | Green | (4,5) |
| *CCNO* | ENSG00000152669 | Green | (6) |
| *CFAP221* | ENSG00000163075 | Red | (7) |
| *CFAP298* | ENSG00000159079 | Green | (4) |
| *CFAP300* | ENSG00000137691 |  | (8,9) |
| *DNAAF1* | ENSG00000154099 | Green | (10) |
| *DNAAF11* | ENSG00000129295 | Green | (11) |
| *DNAAF2* | ENSG00000165506 | Green | (12) |
| *DNAAF3* | ENSG00000167646 | Green | (13) |
| *DNAAF4* | ENSG00000256061 | Green | (14) |
| *DNAAF5* | ENSG00000164818 | Green | (15) |
| *DNAAF6* | ENSG00000080572 | Green | (16,17) |
| *DNAH11* | ENSG00000105877 | Green | (18,19) |
| *DNAH5* | ENSG00000039139 | Green | (20) |
| *DNAH6* | ENSG00000115423 |  | (21) |
| *DNAH8* | ENSG00000124721 | Red | (22) |
| *DNAH9* | ENSG00000007174 | Green | (23–25) |
| *DNAI1* | ENSG00000122735 | Green | (26) |
| *DNAI2* | ENSG00000171595 | Green | (27,28) |
| *DNAJB13* | ENSG00000187726 |  | (29) |
| *DNAL1* | ENSG00000119661 | Green | (30,31) |
| *DRC1* | ENSG00000157856 | Green | (32) |
| *FOXJ1* | ENSG00000129654 |  | (33) |
| *GAS2L2* | ENSG00000270765 | Green | (34) |
| *GAS8* | ENSG00000141013 | Green | (35) |
| *HYDIN* | ENSG00000157423 | Green | (36) |
| *MCIDAS* | ENSG00000234602 | Green | (37) |
| *MNS1* | ENSG00000138587 |  | (38) |
| *NEK10* | ENSG00000163491 | Green | (39) |
| *NME5* | ENSG00000112981 |  | (40,41) |
| *NME8* | ENSG00000086288 | Red | (42) |
| *ODAD1* | ENSG00000105479 | Green | (43,44) |
| *ODAD2P1* | ENSG00000169126 | Green | (45) |
| *ODAD3* | ENSG00000198003 | Green | (46,47) |
| *ODAD4* | ENSG00000204815 |  | (48) |
| *OFD1* | ENSG00000046651 | Red | (49) |
| *RPGR* | ENSG00000156313 | Green | (50) |
| *RSPH1* | ENSG00000160188 | Green | (51) |
| *RSPH3* | ENSG00000130363 | Green | (52) |
| *RSPH4A* | ENSG00000111834 | Green | (53) |
| *RSPH9* | ENSG00000172426 | Green | (53) |
| *SPAG1* | ENSG00000104450 | Green | (54) |
| *SPEF2* | ENSG00000152582 |  | (55,56) |
| *STK36* | ENSG00000163482 |  | (57,58) |
| *TTC12* | ENSG00000149292 | Green | (59) |
| *ZMYND10* | ENSG00000004838 | Green | (60,61) |

Listed are the genes associated with Primary Ciliary Dyskinesia. Genes were identified in the literature, and in the Genomics England Primary Ciliary disorders gene panel (version 1.40) (62). The RAG ratings are as follows green is high evidence, and red is low evidence.

**Supplementary table 2: Optimal ALI time-point (in days) for RNA isolation for known PCD associated genes.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **Ensembl gene identified** | **Time-point** | **Gene** | **Ensembl gene identified** | **Time-point** |
| CCDC103 | ENSG00000167131 | 21 | FOXJ1 | ENSG00000129654 | 21 |
| CCDC39 | ENSG00000284862 | 28 | GAS2L2 | ENSG00000270765 | 21 |
| CCDC40 | ENSG00000141519 | 21 | GAS8 | ENSG00000141013 | 21 |
| CCDC65 | ENSG00000139537 | 21 | HYDIN | ENSG00000157423 | 21 |
| CCNO | ENSG00000152669 | 14 | MCIDAS | ENSG00000234602 | 14 |
| CFAP221 | ENSG00000163075 | 21 | MNS1 | ENSG00000138587 | 21 |
| CFAP298 | ENSG00000159079 | 21 | NEK10 | ENSG00000163491 | 21 |
| CFAP300 | ENSG00000137691 | 21 | NME5 | ENSG00000112981 | 21 |
| DNAAF1 | ENSG00000154099 | 21 | NME8 | ENSG00000086288 | N.A. |
| DNAAF11 | ENSG00000129295 | 21 | ODAD1 | ENSG00000105479 | 21 |
| DNAAF2 | ENSG00000165506 | 21 | ODAD2P1 | ENSG00000238021 | 21 |
| DNAAF3 | ENSG00000167646 | 21 | ODAD3 | ENSG00000198003 | 21 |
| DNAAF4 | ENSG00000256061 | 21 | ODAD4 | ENSG00000204815 | 21 |
| DNAAF5 | ENSG00000164818 | 21 | OFD1 | ENSG00000046651 | 21 |
| DNAAF6 | ENSG00000080572 | 21 | RPGR | ENSG00000156313 | 21 |
| DNAH11 | ENSG00000105877 | 21 | RSPH1 | ENSG00000160188 | 21 |
| DNAH5 | ENSG00000039139 | 21 | RSPH3 | ENSG00000130363 | 21 |
| DNAH6 | ENSG00000115423 | 21 | RSPH4A | ENSG00000111834 | 21 |
| DNAH8 | ENSG00000124721 | 21\* | RSPH9 | ENSG00000172426 | 21 |
| DNAH9 | ENSG00000007174 | 21 | SPAG1 | ENSG00000104450 | 14 |
| DNAI1 | ENSG00000122735 | 21 | SPEF2 | ENSG00000152582 | 21 |
| DNAI2 | ENSG00000171595 | 21 | STK36 | ENSG00000163482 | 21 |
| DNAJB13 | ENSG00000187726 | 21 | TTC12 | ENSG00000149292 | 21 |
| DNAL1 | ENSG00000119661 | 21 | ZMYND10 | ENSG00000004838 | 21 |
| DRC1 | ENSG00000157856 | 21 |  |  |  |

The gene expression of PCD associated genes in eight non-PCD patients was visually assessed to determine the optimal time-point for RNA isolation. Optimal time-points were identified as the time-point with the highest expression together with a low variability. \* very low expression in ALI culture samples (highest median TPM of 1.9x10-03 on ALI-culture day 21 and day 28).

**Supplementary table 3: SpliceAI prediction scores and positions.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Patient** | **Gene** | **Variant** | **Type** | **Score** | **Position** |
| A | *RSPH4A* | c.460C>T | N/A | N/A | N/A |
| B | *DNAH5* | c.10815delT | N/A | N/A | N/A |
|  | *DNAH5* | c.6070\_6071del | Acceptor loss\* | 0.18 | 10 bp |
| C | *HYDIN* | c.2998C>T | Donor loss\* | 0.28 | -44 bp |
|  | *HYDIN* | c.14364delCT | N/A | N/A | N/A |
| D | *HYDIN* | c.2998C>T | Donor loss\* | 0.28 | -44 bp |
|  | *HYDIN* | c.14364delCT | N/A | N/A | N/A |
| E | *DNAH11* | c.983-1G>T | Acceptor loss  Donor loss  Acceptor gain\* | 0.99  0.48  0.64 | 1 bp  212 bp  13 bp |
| F | *CCDC39* | c.357+1G>C | Donor loss  Donor gain | 0.97  0.13 | 1 bp  12 bp |
|  | *CCDC39* | c.664G>T | Acceptor loss  Acceptor gain | 0.41  0.26 | 54 bp  -74 bp |
| 1 | *DNAH11* | c.1974-3C>T | Acceptor loss  Donor loss | 0.50  0.13 | 3 bp  198 bp |
|  | *DNAH11* | c.1741A>T | Acceptor loss | 0.27 | -30 bp |
| 2 | *HYDIN* | c.8973delT | N/A | N/A | N/A |
| 3 | *HYDIN* | c.3786-1G>T | Acceptor loss  Donor loss  Acceptor gain | 0.97  0.79  0.11 | -1 bp  -79 bp  93 bp |
| 4 | *CCDC40* | c.736\_755dup | N/A | N/A | N/A |
|  | *CCDC40* | c.1441-919G>A | Acceptor gain  Donor gain | 0.11  0.23 | -424 bp  1 bp |

The SpliceAI lookup website (https://spliceailookup.broadinstitute.org/) was used to assess the impact of the known genetic variants on splicing using the MANE select transcript. SpliceAI prediction scores of <0.10 are indicated by a N/A. \* Upon visual analysis of splicing in IGV no evidence was seen for the predicted AS.

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