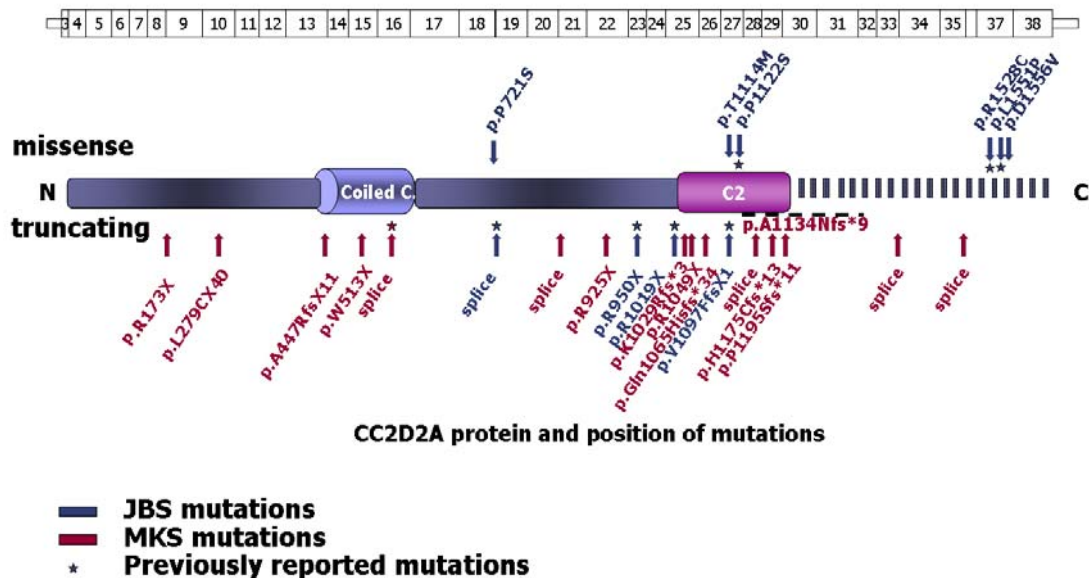


Supp. Table S1. Primers

	Exon	PCR size
CC2D2A-3F : GGTTCCTGGTGAGTGGAAAG CC2D2A-3R : TGGTTGGCCTAACTTCCTCA	3	285
CC2D2A-4F : GCCGCACAGATTGTTAACTG CC2D2A-4R : GAGCAGGCCAAAAGTTGGTA	4	357
CC2D2A-4BF: CGGGGGTCCATACTTAGAGC CC2D2A-4BR: CTTCCTGCCTCCCTTCTC	4B	357
CC2D2A-5F : CCTCTTCCCTGGTGGGTACT CC2D2A-5R : TGGCAATGCATGAAAAATAA	5	399
CC2D2A-6F : GGGGGAGGGAATTGTTTTA CC2D2A-7R : AAGCATTTAATACCTCCTCTGTGC	6-7	693
CC2D2A-8F : GAAATTTGCCAGCATTCCAT1 CC2D2A-8R : CTGTCTTGGCCTCCCAAAG	8	382
CC2D2A-9F: CTCATTAGCATTAAAGCCAGC CC2D2A-9R: AGGGTCTAACCCAGAGCTAC	9	453
CC2D2A-10F: AAAACCCACTTGTAAATATGAAAGG CC2D2A-10R: TGTTTGTGTTGAAGATATTTCTTGATTG	10	482
CC2D2A-11F: GCACATTCATCTTTTCCTTGG CC2D2A-11R: TGCAATGACTGCCCTGTTAG	11	381
CC2D2A-12F: GGGGGAGGAAGTACATGAAA CC2D2A-12R: GCCTGATGTTTGTCTTTATGTGT	12	399
CC2D2A-13F: tggcctctaaaacctggaaa CC2D2A-13R: ctggttagggcgagtgtgtg	13	674
CC2D2A-14F: ctggagacagccagatgctt CC2D2A-14R: cagaagcggtagctgtagca	14	489
CC2D2A-15F: ggttgacatcctattgtcttaca CC2D2A-15R: tttggggattgaggagaatg	15	493
CC2D2A-16F: tgtgcctctgtcatctcatgt CC2D2A-16R: cagagggagccacaggatac	16	468
CC2D2A-17F: tgacctgcctgattacacactt CC2D2A-17R: cctgatgctcgtgtaggtca	17	495
CC2D2A-18F: acctgtggctcctctgggta CC2D2A-18R: ctggcctcccaaagtagca	18	394
CC2D2A-19F: ccacaatgagatgagccctta CC2D2A-19R: ttctatggttgtctgggaagtg	19	377
CC2D2A-20F: gctgtcagcatcatcacctg CC2D2A-20R: tcttgaaaatactggcacctga	20	445
CC2D2A-21F: tccttatgtttaatagccatcatca CC2D2A-21R: tgcataaggagaagatagtgtaaagga	21	398
CC2D2A-22F: ttgtttggaaaatgatttggg CC2D2A-22R: tcaagaaaaagcatggaaagg	22	569
CC2D2A-23F: aaagtttaggaagaaggtggaagaa CC2D2A-23R: ttgaaaggtgatgatagttgtcaat	23	400
CC2D2A-24F: tgcagaggaaagtaacagttgg CC2D2A-24R: tggaaagtagggagggtgaga	24	473
CC2D2A-25F: cagtaggatctcgccattt CC2D2A-25R: aacagtagggccttggcttt	25	489
CC2D2A-26F: gcagtgagctgagattgcac CC2D2A-27R: cgtcaggacacctggatctt	26-27	697
CC2D2A-28F: ccagcagaggggttgtctct CC2D2A-28R: ttgctttgactgtggactc	28	484
CC2D2A-29F: ctagggccacaaagccatta CC2D2A-29R: ctcccaaagtgtgggatta	29	384
CC2D2A-30 : caatgaaaagaaggcaaggaa CC2D2A-30 : ggaaggctctgtttgtgtagg	29b	482
CC2D2A-31F: ttocatatggctgcttacctg CC2D2A-31R: ctcccaaagtgtgggatta	30	687
CC2D2A-32F: aggatgtttggtgaggatgg CC2D2A-32R: tggctaataatgttgcaattta	31	355

	Exon	PCR size
CC2D2A-33F: gagaaaacacccatgccact	32	420
CC2D2A-33R: tttcctagcaacccaaagga		
CC2D2A-34F: gggtgagtaagatgatgactgtcc	33	446
CC2D2A-34R: tcagttatttaaagaacgacagc		
CC2D2A-35F: ggtacacccccaacttcaga	34	466
CC2D2A-35R: gagttgagctcagatgcgttt		
CC2D2A-36F: cttttgggtgggtcatgggta	35	380
CC2D2A-36R: ctttttgagccatgggatt		
CC2D2A-37F: cccagggagtagccattat	36	454
CC2D2A-37R: gtacttctgggagcaacgtg		
CC2D2A-38F: tcttaattgcatacatttcagtagga	37	372
CC2D2A-38R: TGCCAATAATGTTCTTCTGATG		



Supp. Figure S1. CC2D2A protein and position of mutations.

The 38 exons of *CC2D2A* gene encode a protein with predicted Coiled Coil (Coiled C) and C2 domains. Missense and truncating mutations are localised along the entire protein. All truncating mutations result in lethal Meckel syndrome (15/15). By contrast, all but two patients with JS and *CC2D2A* mutations reported here and in previous studies (Gorden et al., 2008; Noor et al., 2008) have at least one missense mutation (10/12) indicating a genotype phenotype correlation.