

Table S2. List of the exome data reanalysis articles and outcome.

No.	No. of undiagnosed exome case	No. of new diagnosis	No. of diagnosed dominant cases (include somatic variants)	Mitochondria variant	CNV	Digenic cases	No. of diagnosed AR cases	No. of homozygous	No. of hemizyous	No. of compound heterozygous	No. of identifying second one in compound heterozygous	Year	PMID
1	40	4	4	0	0	0	0	0	0	0	0	2017	27441994
2	74	27	13	0	0	2	12	6	0	6	0	2017	28327206
3	6	5	3	0	0	0	2	0	1	1	0	2017	27899421
4	33	17	0	0	0	0	17	17	0	0	0	2017	27711071
5	127	14	NA	NA	16	NA	NA	NA	NA	NA	NA	2018	29652076
6	26	4	3	0	0	0	1	1	0	0	0	2018	29595814
7	108	10	4	0	0	0	6	5	1	0	0	2018	29565419
8	156	24	13	0	1	0	10	5	1	4	0	2018	29095811
9	822	182	NA	NA	50	NA	NA	NA	NA	NA	NA	2018	29323667
10	50	6	1	0	0	0	5	4	1	0	0	2018	30125339
11	1684	290	NA	NA	16	NA	NA	NA	NA	NA	1	2019	31216405
12	74	6	6	0	0	0	0	0	0	0	0	2019	30979967
13	139	8	8	0	0	0	0	0	0	0	0	2019	30951195
14	313	48	34	0	0	0	14	9	2	3	0	2019	31231135
15	76	8	7	0	0	0	1	0	0	1	0	2019	30904718
16	83	4	2	0	0	0	2	0	0	2	0	2019	31847883
17	84	13	9	0	0	0	4	3	0	1	0	2019	30377382
18	35	9	6	0	1	0	2	1	0	1	0	2019	29907797
19	2	2	0	0	0	0	2	2	0	0	0	2019	29709712
20	240	38	26	0	0	0	12	9	1	2	0	2019	30577886
21	101	13	8	0	0	0	5	3	1	1	0	2019	30665423
22	46	12	9	0	0	0	3	1	0	2	1	2020	32963807
23	145	18	16	0	0	0	2	1	0	1	0	2020	33303739
24	60	5	1	1	1	0	2	1	0	1	0	2020	31692161
25	58	0	0	0	0	0	0	0	0	0	0	2020	32969205
26	174	14	2	0	4	1	7	3	1	3	1	2021	34560057
27	80	2	0	0	0	0	2	2	0	0	0	2021	33619735
28	20	3	1	0	0	0	2	0	0	2	0	2021	34582790
29	39	2	2	0	0	0	0	0	0	0	0	2021	33962052
30	34**	8	8	0	0	0	0	0	0	0	0	2021	34673242
31	4411	120	69	1	0	0	50	43	3	4	NA	2021	34075210
32	80	7	6	0	0	0	1	1	0	0	0	2022	34906498
33	16243*	119	117	0	0	0	2	1	0	1	0	2022	34906496
34	103	31	23	0	0	0	8	3	3	2	0	2022	35710456
35	323***	67	23	0	0	0	44	22	2	20	NA	2022	35569879
36	150***	48	29	2	1	0	16	9	0	7	NA	2022	34740920
Total	9489	1188					181				3		

*Undiagnosed exome number not clear; **Undiagnosed exome and panel sequencing data; ***Undiagnosed exome, genome and panel sequencing data; NA: not available.