CORRECTION OPEN

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Correction: Structural variant calling and clinical interpretation in 6224 unsolved rare disease exomes

German Demidov, Steven Laurie, Annalaura Torella, Giulio Piluso, Marcello Scala, Manuela Morleo, Vincenzo Nigro, Holm Graessner, Siddharth Banka, Solve-RD consortium, Katja Lohmann and Stephan Ossowski

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Correction to: *European Journal of Human Genetics* (2024) **32**:998–1004 https://doi.org/10.1038/s41431-024-01637-4, published online 31 May 2024

In the original article, Tables 1 and 2 were swapped during initial production.

The incorrectly assigned table order can be seen below:

Table 1. The total number of structural variant calls, the number of evaluated calls, and the diagnostic value increase per ERN. Total number of affected individuals denotes all the affected family members including index cases.

SIMPLE CNVS								
ERN	Chr	Start	End	Manta Type	IGV evaluation	Gene(s)	CNV analysis	Length
GENTURIS	16	68845794	68999240	BND_0/1	DEL	*CDH1*	YES	153446
NMD	10	69917991	69950535	BND_0/1	TANDEM_DUP (within gene)	*MYPN*	YES	32544
ITHACA	6	31630177	31657902	BND_0/1	DEL	*CSNK2B*	YES	27725
NMD	2	179448847	179459005	BND_0/1	DEL	*TTN*	YES	10158
RND	12	58014702	58024265	BND_0/1	DEL	*B4GALNT1*	YES	9563
RND	16	89610020	89619318	BND_1/1	DEL	*SPG7*	YES	9298
ITHACA	2	162269403	162274096	BND_0/1	DEL	*TBR1*	YES	4693
ITHACA	22	51160830	51163907	DEL_0/1	DEL	*SHANK3*	NO	3077
RND	11	66472730	66475186	BND_0/1	DEL	*SPTBN2*	YES	2456
RND	3	38938611	38939384	BND_0/1	DEL	*SCN11A*	NO	773
NMD	17	48247360	48247704	DEL_1/1	DEL	*SGCA*	YES	344
ITHACA	6	1.58E+08	1.58E+08	BND_0/1	DEL	*ARID1B*	NO	245
NMD	6	152485331	152485416	DUP_0/1	TANDEM_DUP (within gene)	*SYNE1*	NO	85
ITHACA	X	73749063	73749133	DUP_0/1	TANDEM_DUP (within gene)	*SLC16A2*	NO	70
ITHACA	14	36987112	36987178	DEL_0/1	DEL	*NKX2-1*	NO	66
PARTS OF COMPLEX EVENTS								
RND	14	50878079	51132277	BND_0/1	INVERSION	*ATL1*	YES	254198
RND	X	153630035	153668349	BND_0/1	COMPLEX_DUP	*ATP6AP1;GDI1;RPL10;TAZ*	YES	38314
ITHACA	16	2214362	2229919	BND_0/1	DUP (partial)	*TRAF7*	YES	15557
RND	3	11070530	11075542	BND_0/1	TANDEM_DUP (within gene) plus DEL	*SLC6A1*	YES	5012
NMD	2	179553482	179557174	DUP_0/1	DEL+DUP	*TTN*	YES	3692
ITHACA	X	152958716	152959469	Mix	Retroduplication	*SLC6A8*	YES	753
COPY-NUMBER NEUTRAL								
ITHACA	9	130887682	140727114	BND_0/1	INVERSION	*EHMT1*	NO	9839432
NMD	X	30960717	31140070	BND_1/1	INVERSION	*DMD*	NO	179353

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Table 2. Variants detected via paired-end or soft-clipped signal based SV analysis (Manta) in exomes, considered to be causative for the corresponding rare diseases.

	ERN RND	ERN ITHACA	ERN NMD	ERN GENTURIS
Number of affected individuals	2.343	1.892	1.632	357
Number of index patients	2.2	1.821	1.499	340
Known disease genes in gene list	1.82	3.081	611	230
Number of candidate variants, after filtering	798	1.404	1.519	15
Number of samples with SVs, after filtering	487	868	606	15
Number of solved index patients/all affected patients	7 (0.32%)/11	9 (0.49%)/9	6 (0.4%)/9	1 (0.29%)/3
Percentage of causal SVs among investigated SVs	1.37%	0.64%	0.59%	20%

The correct table order should have been:

Table 1. Table 1. The total number of structural variant calls, the number of evaluated calls, and the diagnostic value increase per ERN. Total number of affected individuals denotes all the affected family members including index cases.

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The original article has now been corrected.