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Whole exome sequencing studies in epilepsy: a deep analysis of the published literature

AUTHOR(S)

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CITATION

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WES item	Item number	Relative Risk of Genetics vs. Neurology journal (95% CI)	Relative Risk of Neurology vs. other journal (95% CI)	Relative Risk of Genetics vs. other journal (95% CI)	Relative Risk of Post 2016 vs. Pre 2017 (95% CI)	Relative Risk of High impact factor vs. Low impact factor journals (95% CI)	Relative Risk of Cases vs. Cohort (95% CI)	Percentage of total number of studies meeting item (%)
Title mentions gene	1	1.03 (0.95-1.12)	0.95 (0.85-1.06)	0.98 (0.88-1.08)	0.92 (0.86-0.99)	1.10 (0.92-1.12)	1.39 (1.20-1.62)	87
Abstract mentions variant	2	0.98 (0.84-1.14)	0.95 (0.76-1.18)	0.93 (0.75-1.14)	0.98 (0.85-1.12)	0.79 (0.62-0.99)	2.22 (1.63-3.02)	63
Abstract mentions gene	3	1.00 (0.96-1.03)	1.01 (0.95-1.06)	1.00 (0.95-1.06)	0.98 (0.95-1.02)	0.99 (0.95-1.04)	1.11 (1.03-1.20)	97
Abstract mentions syndrome	4	0.99 (0.95-1.02)	1.05 (0.97-1.13)	1.04 (0.96-1.12)	0.98 (0.95-1.02)	0.99 (0.94-1.04)	1.06 (1.03-1.12)	96
Clinical features	5	0.96 (0.92-1.0)	1.02 (0.96-1.09)	0.99 (0.92-1.06)	1.00 (0.96-1.04)	0.89 (0.81-0.97)	1.13 (1.04-1.24)	95
Ethnicity	6	1.11 (0.94-1.30)	0.99 (0.77-1.28)	1.10 (0.86-1.40)	0.97 (0.84-1.13)	1.00 (0.81-1.24)	1.18 (0.96-1.46)	61
Human Phenotype terms	7	8.62 (1.08-63.37)	1.02 (0.04-24.78)	5.39 (0.32-90.63)	1.09 (0.37-3.20)	0.71 (0.15-3.33)	0.57 (0.18-1.81)	3
OMIM	8	1.88 (1.40-2.53)	1.09 (0.63-1.88)	2.05 (1.24-3.37)	0.87 (0.68-1.11)	1.47 (1.09-1.98)	0.71 (0.54-0.93)	35
Consanguinity	9	1.12 (0.94-1.32)	1.22 (0.89-1.67)	1.36 (1.01-1.84)	0.88 (0.75-1.03)	1.09 (0.88-1.37)	1.74 (1.31-2.31)	58
Inclusion/selection of patients	10	0.56 (0.28-1.13)	1.15 (0.95-1.39)	0.64 (0.32-1.31)	1.17 (0.93-1.46)	0.87 (0.64-1.17)	Not estimable	80
Ancillary Investigations	11	1.02 (0.95-1.10)	1.00 (0.89-1.12)	1.02 (0.91-1.13)	0.97 (0.91-1.04)	0.97 (0.87-1.07)	1.15 (1.03-1.28)	88
Prior genetic tests	12	1.24 (0.94-1.63)	1.40 (0.83-2.37)	1.73 (1.05-2.87)	1.04 (0.80-1.35)	1.28 (0.89-1.84)	0.72 (0.54-0.94)	35
Pedigree chart	13	1.18 (0.97-1.43)	0.91 (0.68-1.22)	1.07 (0.82-1.41)	0.87 (0.73-1.04)	0.90 (0.68-1.18)	1.69 (1.25-2.28)	53
Molecular science background	14	1.07 (0.96-1.19)	1.08 (0.89-1.30)	1.15 (0.96-1.38)	1.19 (1.08-1.32)	1.11 (0.98-1.27)	1.01 (0.89-1.15)	78

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DNA source	15	1.23 (0.98-1.55)	0.66 (0.50-0.88)	0.82 (0.62-1.05)	1.16 (0.95-1.42)	0.82 (0.61-1.12)	1.05 (0.81-1.35)	47
Exome target kit	16	1.26 (1.09- 1.45)	1.03 (0.80-1.33)	1.30 (1.03-1.64)	0.85 (0.74-0.96)	1.18 (0.98-1.42)	0.91 (0.78-1.05)	67
Sequencer used	17	1.14 (1.01-1.29)	1.10 (0.88-1.37)	1.25 (1.02-1.54)	0.90 (0.81-1.01)	1.13 (0.97-1.32)	0.89 (0.79-1.01)	73
Quality scores	18	1.95 (1.11-3.42)	0.46 (0.22-0.94)	0.90 (0.49-1.64)	1.24 (0.78-1.95)	1.10 (0.59-2.06)	0.92 (0.53-1.59)	14
Reference genome	19	1.57 (1.23-1.99)	0.93 (0.63-1.39)	1.46 (1.02-2.09)	1.23 (1.00-1.51)	1.39 (1.09-1.77)	0.93 (0.73-1.19)	45
Haplotype caller	20	1.24 (1.04-1.47)	0.99 (0.74-1.32)	1.22 (0.94-1.60)	1.04 (0.89-1.21)	1.30 (1.06-1.59)	0.79 (0.67-0.92)	58
Variant filtering	21	1.26 (1.02-1.55)	1.17 (0.80-1.72)	1.47 (1.03-2.11)	1.18 (0.97-1.43)	1.50 (1.18-1.72)	0.80 (0.65-0.99)	48
Annotation database	22	1.21 (0.94-1.55)	1.08 (0.71-1.65)	1.31 (0.88-1.94)	1.20 (0.95-1.51)	1.40 (1.04-1.89)	0.77 (0.60-0.99)	38
Minor allele frequency	23	1.19 (1.04-1.47)	1.09 (0.86-1.39)	1.30 (1.04-1.62)	1.12 (0.99-1.26)	1.15 (0.98-1.34)	1.00 (0.86-1.16)	71
In-silico predicting databases	24	1.12 (0.95-1.33)	1.19 (0.88-1.61)	1.35 (1.01-1.81)	1.19 (1.02-1.39)	0.92 (0.73-1.16)	1.04 (0.85-1.27)	59
Evolution conservation databases	25	1.54 (1.12-2.12)	0.67 (0.43-1.05)	1.04 (0.7-1.54)	0.99 (0.75-1.30)	1.0 (0.68-1.46)	1.03 (0.73-1.45)	31
How variant impacts polypeptide	26	1.32 (1.13-1.55)	0.91 (0.71-1.18)	1.21 (0.96-1.52)	0.80 (0.70-0.92)	1.72 (1.44-2.05)	1.15 (0.95-1.39)	64
Pathogenic database of previous reports	27	1.12 (0.91-1.39)	1.01 (0.72-1.42)	1.13 (0.82-1.57)	1.29 (1.06-1.58)	0.87 (0.63-1.20)	1.32 (0.99-1.76)	47
ACMG guidelines applied	28	1.88 (1.04-3.38)	1.01 (0.72-1.42)	0.99 (0.50-1.93)	12.63 (4.65-34.29)	0.59 (0.25-1.39)	0.24 (0.15-0.37)	13

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Diagnostic yield estimate	29	1.01 (0.60-1.73)	0.84 (0.34-2.07)	0.53 (0.31-0.91)	1.10 (0.67-1.79)	0.50 (0.26-0.95)	Not estimable	43
Sanger sequencing	30	1.14 (1.02-1.28)	0.95 (0.80-1.13)	1.08 (0.92-1.27)	0.90 (0.82-1.00)	1.01 (0.87-1.16)	0.97 (0.86-1.09)	77
Full transcript information	31	1.41 (1.13-1.76)	1.08 (0.73-1.59)	1.52 (1.06-2.18)	1.45 (1.18-1.77)	1.51 (1.23-1.84)	1.10 (0.85-1.42)	47
VUS reported	32	1.05 (0.69-1.61)	0.78 (0.44-1.39)	0.82 (0.47-1.42)	1.00 (0.68-1.47)	1.12 (0.63-1.98)	0.57 (0.38-0.86)	19
Mutation type	33	1.04 (0.98-1.09)	0.94 (0.89-0.99)	0.97 (0.93-1.02)	0.97 (0.93-1.01)	1.04 (0.98-1.10)	1.00 (0.95-1.06)	95
Variant segregation/inheritance	34	0.98 (0.92-1.05)	0.99 (0.91-1.07)	0.97 (0.89-1.05)	0.97 (0.91-1.03)	1.00 (0.92-1.09)	0.98 (0.92-1.05)	91
Sequence chronogram	35	1.67 (1.34-2.07)	0.62 (0.47-0.82)	1.03 (0.82-1.29)	1.02 (0.83-1.25)	0.76 (0.54-1.07)	1.98 (1.37-2.86)	45
Previous literature	36	1.01 (0.99-1.03)	1.60 (1.30-1.97)	1.03 (0.98-1.08)	0.99 (0.97-1.01)	0.98 (0.95-1.02)	1.00 (0.96-1.04)	98
Deposit findings to ClinVar	37	2.50 (1.03-6.10)	0.67 (0.17-2.61)	1.69 (0.52-5.49)	2.90 (1.28-6.75)	1.71 (0.76-3.84)	0.56 (0.26-1.19)	6
Phenotype compared to reported associated disease	38	0.98 (0.91-1.05)	1.21 (1.03-1.43)	1.19 (1.01-1.40)	0.97 (0.91-1.05)	0.75 (0.58-0.98)	1.16 (1.03-1.30)	86

