

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

AUTHOR(S)

Arif Shukralla, Robert Carton, Katherine Benson, Hany El-Naggar, Austin Lacey, Gianpiero Cavalleri, Norman Delanty

CITATION

Shukralla, Arif; Carton, Robert; Benson, Katherine; El-Naggar, Hany; Lacey, Austin; Cavalleri, Gianpiero; et al. (2022): Whole exome sequencing studies in epilepsy: a deep analysis of the published literature. Royal College of Surgeons in Ireland. Journal contribution. <https://hdl.handle.net/10779/rcsi.19267880.v1>

HANDLE

[10779/rcsi.19267880.v1](https://hdl.handle.net/10779/rcsi.19267880.v1)

LICENCE

CC BY-NC-SA 4.0

This work is made available under the above open licence by RCSI and has been printed from <https://repository.rcsi.com>. For more information please contact repository@rcsi.com

URL

https://repository.rcsi.com/articles/journal_contribution/Whole_exome_sequencing_studies_in_epilepsy_a_deep_analysis_of_the_published_literature/19267880/1

WES item	Item number	High Impact Genetics Journals vs. Low impact Genetics Journals	High Impact Genetics Journals vs. High Impact Neurology Journals
Title mentions gene	1	0.99 (0.87-1.12)	1.03 (0.86-1.24)
Abstract mentions variant	2	1.44 (1.02-2.02)	0.88 (0.57-1.35)
Abstract mentions gene	3	1.00 (0.94-1.06)	0.99 (0.90-1.09)
Abstract mentions syndrome	4	0.98 (0.93-1.03)	0.94 (0.85-1.03)
Clinical features	5	1.18 (1.03-1.35)	0.94 (0.79-1.13)
Ethnicity	6	0.94 (0.72-1.23)	2.39 (1.28-4.43)
Human Phenotype number	7	2.79 (0.34-23.13)	0.64 (0.04-9.77)
OMIM	8	0.69 (0.51-0.95)	2.39 (1.28-4.43)
Consanguinity	9	0.88 (0.66-1.18)	1.19 (0.82-1.74)
Inclusion/selection of patients	10	2.10 (1.09-4.06)	0.73 (0.50-1.05)
Ancillary Investigations	11	1.06 (0.92-1.21)	1.01 (0.83-1.22)
Prior genetic tests	12	0.71 (0.44-1.13)	1.59 (0.81-3.11)
Pedigree chart	13	1.14 (0.79-1.65)	1.17 (0.69-1.96)
Molecular science background	14	0.90 (0.75-1.08)	1.07 (0.85-1.35)

WES item	Item number	High Impact Genetics Journals vs. Low impact Genetics Journals	High Impact Genetics Journals vs. High Impact Neurology Journals
DNA source	15	1.45 (0.95-2.19)	1.15 (0.62-2.11)
Exome target kit	16	0.99 (0.77-1.26)	0.81 (0.65-1.02)
Sequencer used	17	1.03 (0.84-1.26)	0.87 (0.70-1.06)
Quality control	18	0.68 (0.31-1.52)	1.91 (0.56-6.45)
Reference genome	19	0.99 (0.73-1.34)	0.94 (0.67-1.32)
Haplotype caller	20	0.86 (0.67-1.09)	1.03 (0.79-1.35)
Variant filtering	21	0.92 (0.68-1.26)	0.95 (0.66-1.37)
Annotation database	22	0.88 (0.59-1.31)	0.93 (0.59-1.47)
Minor allele frequency	23	1.00 (0.83-1.21)	1.06 (0.82-1.38)
<i>In-silico</i> predicting databases	24	1.26 (0.92-1.74)	0.95 (0.63-1.45)
Evolution conservation databases	25	0.99 (0.62-1.57)	1.64 (0.79-3.41)
How variant impacts polypeptide	26	0.77 (0.61-0.96)	0.92 (0.76-1.11)
Pathogenic database of previous reports	27	1.06 (0.70-1.60)	1.41 (0.75-2.65)
ACMG guidelines applied	28	2.09 (0.74-5.91)	3.18 (0.39-25.83)

WES item	Item number	High Impact Genetics Journals vs. Low impact Genetics Journals	High Impact Genetics Journals vs. High Impact Neurology Journals
Diagnostic yield estimate	29	0.94 (0.17-5.30)	2.33 (0.58-9.38)
Sanger sequencing	30	1.08 (0.91-1.30)	1.06 (0.82-1.38)
Full transcript information	31	0.95 (0.68-1.34)	1.59 (0.91-2.78)
VUS reported	32	1.04 (0.45-2.40)	0.64 (0.25-1.62)
Mutation type	33	1.01 (0.93-1.09)	0.99 (0.90-1.09)
Variant segregation/inheritance	34	1.13 (0.98-1.30)	0.85 (0.74-0.97)
Sequence chronogram	35	1.27 (0.78-2.04)	0.95 (0.50-1.82)
Previous literature	36	1.03 (0.97-1.09)	0.98 (0.91-1.06)
Deposit findings to ClinVar	37	0.65 (0.23-1.82)	3.82 (0.49-30.06)
Phenotype compared to reported associated disease	38	1.17 (0.99-1.38)	0.89 (0.73-1.09)