Louise E Bier

List of Publications by Year
in descending order

16 papers	858 citations	933447 10 h-index	940533 16 g-index
17 all docs	17 docs citations	17 times ranked	1716 citing authors

#	Article	IF	Citations
1	Whole-exome sequencing in the evaluation of fetal structural anomalies: a prospective cohort study. Lancet, The, 2019, 393, 758-767.	13.7	368
2	Comparison of Parkinson Risk in Ashkenazi Jewish Patients With Gaucher Disease andGBAHeterozygotes. JAMA Neurology, 2014, 71, 752.	9.0	172
3	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	3.9	60
4	Genetic testing for the epilepsies: A systematic review. Epilepsia, 2022, 63, 375-387.	5.1	53
5	Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. American Journal of Medical Genetics, Part A, 2018, 176, 2259-2275.	1.2	47
6	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
7	Early manifestations of type 1 Gaucher disease in presymptomatic children diagnosed after parental carrier screening. Genetics in Medicine, 2017, 19, 652-658.	2.4	19
8	Genetic testing in individuals with cerebral palsy. Developmental Medicine and Child Neurology, 2021, 63, 1448-1455.	2.1	19
9	Lossâ€ofâ€function variants in NFIA provide further support that NFIA is a critical gene in 1p32â€p31 deletion syndrome: A four patient series. American Journal of Medical Genetics, Part A, 2017, 173, 3158-3164.	1.2	16
10	Patientsâ€™ Opinions on Genetic Counseling on the Increased Risk of Parkinson Disease among Gaucher Disease Carriers. Journal of Genetic Counseling, 2018, 27, 675-680.	1.6	14
11	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
12	Evaluation of the cost and effectiveness of diverse recruitment methods for a genetic screening study. Genetics in Medicine, 2019, 21, 2371-2380.	2.4	10
13	A Novel Kv7.3 Variant in the Voltage-Sensing S4 Segment in a Family With Benign Neonatal Epilepsy: Functional Characterization and in vitro Rescue by I²-Hydroxybutyrate. Frontiers in Physiology, 2020, 11, 1040.	2.8	7
14	Cases in Precision Medicine: Genetic Testing to Predict Future Risk for Disease in a Healthy Patient. Annals of Internal Medicine, 2021, 174, 540-547.	3.9	7
15	Truncating variants in the SHANK1 gene are associated with a spectrum of neurodevelopmental disorders. Genetics in Medicine, 2021, 23, 1912-1921.	2.4	5
16	Diagnostic sequencing to support genetically stratified medicine in a tertiary care setting. Genetics in Medicine, 2022, 24, 862-869.	2.4	4