Deborah Lambert

List of Publications by Year
in descending order

27 papers	1,589 citations	687363 13 h-index	642732 23 g-index
31 all docs	31 docs citations	31 times ranked	2358 citing authors

#	Article	IF	Citations
1	Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. European Journal of Human Genetics, 2020, 28, 165-173.	2.8	716
2	The Global State of the Genetic Counseling Profession. European Journal of Human Genetics, 2019, 27, 183-197.	2.8	215
3	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	2.5	196
4	Outcomes of Siblings with Classical Galactosemia. Journal of Pediatrics, 2009, 154, 721-726.	1.8	116
5	Keutel syndrome: Further characterization and review. American Journal of Medical Genetics Part A, 1998, 78, 182-187.	2.4	55
6	In Vivo Variability of TMA Oxidation Is Partially Mediated by Polymorphisms of the FMO3 Gene. Molecular Genetics and Metabolism, 2001, 73, 224-229.	1.1	45
7	A profile of the genetic counsellor and genetic nurse profession in European countries. Journal of Community Genetics, 2012, 3, 19-24.	1.2	37
8	Development of a registration system for genetic counsellors and nurses in health-care services in Europe. European Journal of Human Genetics, 2016, 24, 312-314.	2.8	27
9	A study of the practice of individual genetic counsellors and genetic nurses in Europe. Journal of Community Genetics, 2013, 4, 69-75.	1.2	26
10	Short-chain hydroxyacylâ€“coenzyme A dehydrogenase deficiency presenting as unexpected infant death: A family study. Journal of Pediatrics, 2000, 137, 257-259.	1.8	19
11	Catalogue of inherited disorders found among the Irish Traveller population. Journal of Medical Genetics, 2018, 55, 233-239.	3.2	19
12	Polymorphisms of the Flavin containing monooxygenase 3 (FMO3) gene do not predispose to essential hypertension in Caucasians. BMC Medical Genetics, 2005, 6, 41.	2.1	18
13	The role of primary care in management of rare diseases in Ireland. Irish Journal of Medical Science, 2020, 189, 771-776.	1.5	15
14	The stepwise process of integrating a genetic counsellor into primary care. European Journal of Human Genetics, 2022, 30, 772-781.	2.8	15
15	The recognition of the profession of Genetic Counsellors in Europe. European Journal of Human Genetics, 2018, 26, 1719-1720.	2.8	14
16	A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland. Orphanet Journal of Rare Diseases, 2020, 15, 311.	2.7	11
17	Genetic counselors and legal recognition: A madeâ€forâ€Canada approach. Journal of Genetic Counseling, 2022, 31, 49-58.	1.6	9
18	The perceived impact of the European registration system for genetic counsellors and nurses. European Journal of Human Genetics, 2017, 25, 1075-1077.	2.8	8

#	Article	IF	Citations
19	The Camera-Marugo-Cohen syndrome: Report of two new patients. , 1999, 86, 208-214.		7
20	Communication of Genetic Information by Other Health Professionals: The Role of the Genetic Counsellor in Specialist Clinics. Journal of Genetic Counseling, 2011, 20, 192-203.	1.6	7
21	Incidence of Fragile X syndrome in Ireland. American Journal of Medical Genetics, Part A, 2017, 173, 678-683.	1.2	4
22	Not Camera-Marugo-Cohen syndrome but diploid/triploid mixoploidy. American Journal of Medical Genetics Part A, 2001, 104, 343-344.	2.4	3
23	An estimate of the cumulative paediatric prevalence of rare diseases in Ireland and comment on the literature. European Journal of Human Genetics, 2022, 30, 1211-1215.	2.8	3
24	Keutel syndrome: Further characterization and review. American Journal of Medical Genetics Part A, 1998, 78, 182-187.	2.4	1
25	GP165â€...Towards estimating the incidence of rare diseases in a paediatric population, born in ireland in the year 2000. , 2019, , .		0
26	Reply to E. Vicente et al.. European Journal of Human Genetics, 2021, 29, 1034-1035.	2.8	0
27	Fatal fetal abnormality Irish live-born survivalâ€"an observational study. Journal of Community Genetics, 2021, 12, 643-651.	1.2	0