Suzanne C E H Sallevelt

List of Publications by Year
in descending order

15	836	759233 12	940533 16
papers	citations	h-index	g-index

16	16	16	2037
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The expanding phenotype of COL4A1 and COL4A2 mutations: clinical data on 13 newly identified families and a review of the literature. Genetics in Medicine, 2015, 17, 843-853.	2.4	204
2	Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599.	2.8	104
3	Preimplantation genetic diagnosis in mitochondrial DNA disorders: challenge and success. Journal of Medical Genetics, 2013, 50, 125-132.	3.2	81
4	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
5	Whole Exome Sequencing Is the Preferred Strategy to Identify the Genetic Defect in Patients With a Probable or Possible Mitochondrial Cause. Frontiers in Genetics, 2018, 9, 400.	2.3	66
6	Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041.	2.9	53
7	Preventing the transmission of mitochondrial DNA disorders using prenatal or preimplantation genetic diagnosis. Annals of the New York Academy of Sciences, 2015, 1350, 29-36.	3.8	52
8	Rapid whole exome sequencing in pregnancies to identify the underlying genetic cause in fetuses with congenital anomalies detected by ultrasound imaging. Prenatal Diagnosis, 2020, 40, 972-983.	2.3	49
9	Mutation-specific effects in germline transmission of pathogenic mtDNA variants. Human Reproduction, 2018, 33, 1331-1341.	0.9	36
10	A comprehensive strategy for exome-based preconception carrier screening. Genetics in Medicine, 2017, 19, 583-592.	2.4	30
11	Genetic defects in mtDNA-encoded protein translation cause pediatric, mitochondrial cardiomyopathy with early-onset brain disease. European Journal of Human Genetics, 2018, 26, 537-551.	2.8	23
12	Diagnostic exome-based preconception carrier testing in consanguineous couples: results from the first 100 couples in clinical practice. Genetics in Medicine, 2021, 23, 1125-1136.	2.4	20
13	Anatomic & metabolic brain markers of the m.3243A>G mutation: A multi-parametric 7T MRI study. NeuroImage: Clinical, 2018, 18, 231-244.	2.7	15
14	Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect. Frontiers in Molecular Neuroscience, 2017, 10, 336.	2.9	11
15	Pathogenic SLIRP variants as a novel cause of autosomal recessive mitochondrial encephalomyopathy with complex I and IV deficiency. European Journal of Human Genetics, 2021, 29, 1789-1795.	2.8	7