# 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 | 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 |
| 2 | 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 |
| 3 | 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 |
| 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 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | Mutation-specific effects in germline transmission of pathogenic mtDNA variants. Human Reproduction, 2018, 33, 1331-1341. | 0.9 | 36 |
| 9 | Diagnostic exome sequencing in 266 Dutch patients with visual impairment. European Journal of Human Genetics, 2017, 25, 591-599. | 2.8 | 104 |
| 10 | A comprehensive strategy for exome-based preconception carrier screening. Genetics in Medicine, 2017, 19, 583-592. | 2.4 | 30 |
| 11 | Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect. Frontiers in Molecular Neuroscience, 2017, 10, 336. | 2.9 | 11 |
| 12 | Mitochondrial DNA sequence characteristics modulate the size of the genetic bottleneck. Human Molecular Genetics, 2016, 25, 1031-1041. | 2.9 | 53 |
| 13 | 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 |
| 14 | 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 |
| 15 | Preimplantation genetic diagnosis in mitochondrial DNA disorders: challenge and success. Journal of Medical Genetics, 2013, 50, 125-132. | 3.2 | 81 |