Klaske D Lichtenbelt

List of Publications by Year
in descending order

27	1,369	394421 19	552781 26
papers	citations	h-index	g-index

27	27	27	2625
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	TRIDENT-2: National Implementation of Genome-wide Non-invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands. American Journal of Human Genetics, 2019, 105, 1091-1101.	6.2	222
2	Phenotypic variability in patients with ADA2 deficiency due to identical homozygous R169Q mutations. Rheumatology, 2016, 55, 902-910.	1.9	116
3	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	2.5	116
4	Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management. European Journal of Human Genetics, 2015, 23, 1286-1293.	2.8	108
5	Cardiovascular malformations caused by NOTCH1 mutations do not keep left: data on 428 probands with left-sided CHD and their families. Genetics in Medicine, 2016, 18, 914-923.	2.4	104
6	Origin and clinical relevance of chromosomal aberrations other than the common trisomies detected by genome-wide NIPS: results of the TRIDENT study. Genetics in Medicine, 2018, 20, 480-485.	2.4	85
7	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	6.2	71
8	Trial by Dutch laboratories for evaluation of nonâ€invasive prenatal testing. Part IIâ€”women's perspectives. Prenatal Diagnosis, 2016, 36, 1091-1098.	2.3	62
9	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
10	CTCF variants in 39 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum. Genetics in Medicine, 2019, 21, 2723-2733.	2.4	48
11	Women's Experience with Nonâ€Invasive Prenatal Testing and Emotional Wellâ€being and Satisfaction after Testâ€Results. Journal of Genetic Counseling, 2017, 26, 1348-1356.	1.6	42
12	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	2.4	41
13	Clinical impact of additional findings detected by genome-wide non-invasive prenatal testing: Follow-up results of the TRIDENT-2 study. American Journal of Human Genetics, 2022, 109, 1140-1152.	6.2	39
14	Trends in the utilization of invasive prenatal diagnosis in The Netherlands during 2000â€“2009. Prenatal Diagnosis, 2011, 31, 765-772.	2.3	29
15	Clinical presentation and spectrum of neuroimaging findings in newborn infants with incontinentia pigmenti. Developmental Medicine and Child Neurology, 2016, 58, 1076-1084.	2.1	28
16	Preimplantation Genetic Testing for Monogenic Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 1279-1286.	4.5	27
17	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	8.2	27
18	De novo variants in CUL3 are associated with global developmental delays with or without infantile spasms. Journal of Human Genetics, 2020, 65, 727-734.	2.3	23

#	Article	IF	Citations
19	Noninvasive Prenatal Test Results Indicative of Maternal Malignancies: A Nationwide Genetic and Clinical Follow-Up Study. Journal of Clinical Oncology, 2022, 40, 2426-2435.	1.6	23
20	Genomic and Functional Overlap between Somatic and Germline Chromosomal Rearrangements. Cell Reports, 2014, 9, 2001-2010.	6.4	21
21	Factors determining uptake of invasive testing following firstâ€trimester combined testing. Prenatal Diagnosis, 2013, 33, 328-333.	2.3	17
22	Maternal vitamin B12 deficiency and abnormal cell-free DNA results in pregnancy. Prenatal Diagnosis, 2016, 36, 790-793.	2.3	17
23	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. American Journal of Neuroradiology, 2018, 39, 1146-1152.	2.4	12
24	NANS-CDG: Delineation of the Genetic, Biochemical, and Clinical Spectrum. Frontiers in Neurology, 2021, 12, 668640.	2.4	11
25	Accuracy of diagnosis and counseling of fetal brain anomalies prior to 24 weeks of gestational age. Journal of Maternal-Fetal and Neonatal Medicine, 2018, 31, 2188-2194.	1.5	10
26	De novo heterozygous missense and lossâ€ofâ€function variants in CDC42BPB are associated with a neurodevelopmental phenotype. American Journal of Medical Genetics, Part A, 2020, 182, 962-973.	1.2	8
27	P0050PRE-IMPLANTATION GENETIC TESTING FOR MONOGENIC KIDNEY DISEASE: TWENTY-FIVE YEAR EXPERIENCE IN THE NETHERLANDS. Nephrology Dialysis Transplantation, 2020, 35, .	0.7	0