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	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
2	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
3	NANS-CDG: Delineation of the Genetic, Biochemical, and Clinical Spectrum. Frontiers in Neurology, 2021, 12, 668640.	2.4	11
4	P0050PRE-IMPLANTATION GENETIC TESTING FOR MONOGENIC KIDNEY DISEASE: TWENTY-FIVE YEAR EXPERIENCE IN THE NETHERLANDS. Nephrology Dialysis Transplantation, 2020, 35, .	0.7	0
5	Preimplantation Genetic Testing for Monogenic Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 1279-1286.	4.5	27
6	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
7	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
8	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
9	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
10	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
11	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
12	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	8.2	27
13	Characteristic MR Imaging Findings of the Neonatal Brain in RASopathies. American Journal of Neuroradiology, 2018, 39, 1146-1152.	2.4	12
14	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	2.5	116
15	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
16	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
17	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
18	Maternal vitamin B12 deficiency and abnormal cell-free DNA results in pregnancy. Prenatal Diagnosis, 2016, 36, 790-793.	2.3	17

#	Article	IF	Citations
19	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
20	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
21	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
22	Phenotypic variability in patients with ADA2 deficiency due to identical homozygous R169Q mutations. Rheumatology, 2016, 55, 902-910.	1.9	116
23	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
24	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	6.2	71
25	Genomic and Functional Overlap between Somatic and Germline Chromosomal Rearrangements. Cell Reports, 2014, 9, 2001-2010.	6.4	21
26	Factors determining uptake of invasive testing following firstâ€trimester combined testing. Prenatal Diagnosis, 2013, 33, 328-333.	2.3	17
27	Trends in the utilization of invasive prenatal diagnosis in The Netherlands during 2000â€"2009. Prenatal Diagnosis, 2011, 31, 765-772.	2.3	29