Elisa Contini

List of Publications by Year in descending order

11 papers	144 citations	1478505 6 h-index	1281871 11 g-index
11 all docs	11 docs citations	11 times ranked	366 citing authors

#	Article	IF	Citations
1	Defining the diagnostic effectiveness of genes for inclusion in panels: the experience of two decades of genetic testing for hypertrophic cardiomyopathy at a single center. Genetics in Medicine, 2019, 21, 284-292.	2.4	54
2	Mutation landscape in patients with myelofibrosis receiving ruxolitinib or hydroxyurea. Blood Cancer Journal, 2018, 8, 122.	6.2	25
3	Clinical and genetic study of a family with a paternally inherited 15q11â€“q13 duplication. American Journal of Medical Genetics, Part A, 2013, 161, 1459-1464.	1.2	17
4	A Systematic Assessment of Accuracy in Detecting Somatic Mosaic Variants by Deep Amplicon Sequencing: Application to NF2 Gene. PLoS ONE, 2015, 10, e0129099.	2.5	16
5	Characteristics and clinical correlates of NFE2 mutations in chronic Myeloproliferative neoplasms. American Journal of Hematology, 2020, 95, E23-E26.	4.1	8
6	Validation of a method for noninvasive prenatal testing for fetal aneuploidies risk and considerations for its introduction in the Public Health System. Journal of Maternal-Fetal and Neonatal Medicine, 2017, 30, 710-716.	1.5	6
7	Fundus phenotype in retinitis pigmentosa associated with EYS mutations. Ophthalmic Genetics, 2018, 39, 589-602.	1.2	6
8	Growth hormone therapy-related hyperglycaemia in a boy with renal cystic hypodysplasia and a new mutation of the HNF1Â gene. Nephrology Dialysis Transplantation, 2010, 25, 3116-3119.	0.7	4
9	Long Reads, Short Time: Feasibility of Prenatal Sample Karyotyping by Nanopore Genome Sequencing. Clinical Chemistry, 2019, 65, 1605-1608.	3.2	4
10	Mutated ASXL1 and number of somatic mutations as possible indicators of progression to chronic myelomonocytic leukemia of myelodysplastic syndromes with single or multilineage dysplasia. Haematologica, 2017, 102, e332-e335.	3.5	2
11	Germline Mutation in KIF1BÎ² Gene Associated with Loss of Heterozygosity: Usefulness of Next-Generation Sequencing in the Genetic Screening of Patients with Pheochromocytoma. International Journal of Endocrinology, 2020, 2020, 1-8.	1.5	2