Giancarlo Deidda

List of Publications by Year
in descending order

23 papers	1,237 citations	567281 15 h-index	642732 23 g-index
23 all docs	23 docs citations	23 times ranked	978 citing authors

#	Article	IF	Citations
1	Progress in the molecular diagnosis of facioscapulohumeral muscular dystrophy and correlation between the number ofKpnI repeats at the 4q35 locus and clinical phenotype. Annals of Neurology, 1999, 45, 751-757.	5.3	263
2	De Novo Facioscapulohumeral Muscular Dystrophy: Frequent Somatic Mosaicism, Sex-Dependent Phenotype, and the Role of Mitotic Transchromosomal Repeat Interaction between Chromosomes 4 and 10. American Journal of Human Genetics, 2000, 66, 26-35.	6.2	136
3	Direct detection of 4q35 rearrangements implicated in facioscapulohumeral muscular dystrophy (FSHD).. Journal of Medical Genetics, 1996, 33, 361-365.	3.2	129
4	Inter- and intrachromosomal sub-telomeric rearrangements on 4q35: implications for facioscapulohumeral muscular dystrophy (FSHD) aetiology and diagnosis. Human Molecular Genetics, 1998, 7, 1207-1214.	2.9	96
5	Interchromosomal repeat array interactions between chromosomes 4 and 10: a model for subtelomeric plasticity. Human Molecular Genetics, 2000, 9, 2879-2884.	2.9	95
6	Physical Mapping Evidence for a Duplicated Region on Chromosome 10qter Showing High Homology with the Facioscapulohumeral Muscular Dystrophy Locus on Chromosome 4qter. European Journal of Human Genetics, 1995, 3, 155-167.	2.8	89
7	FRG2, an FSHD candidate gene, is transcriptionally upregulated in differentiating primary myoblast cultures of FSHD patients. Journal of Medical Genetics, 2004, 41, 826-836.	3.2	76
8	Sequence Homology between 4qter and 10qter Loci Facilitates the Instability of Subtelomeric KpnI Repeat Units Implicated in Facioscapulohumeral Muscular Dystrophy. American Journal of Human Genetics, 1998, 63, 181-190.	6.2	60
9	Allele-specific DNA hypomethylation characterises FSHD1 and FSHD2. Journal of Medical Genetics, 2016, 53, 348-355.	3.2	54
10	Estrogens enhance myoblast differentiation in facioscapulohumeral muscular dystrophy by antagonizing DUX4 activity. Journal of Clinical Investigation, 2017, 127, 1531-1545.	8.2	46
11	Molecular characterization of Î²-thalassemia mutations in Egypt. Human Genetics, 1990, 85, 272-274.	3.8	39
12	Molecular analysis of 4q35 rearrangements in facioscapulohumeral muscular dystrophy (FSHD): application to family studies for a correct genetic advice and a reliable prenatal diagnosis of the disease. Neuromuscular Disorders, 1999, 9, 190-198.	0.6	26
13	An archaeal endoribonuclease catalyzes cis- and trans- nonspliceosomal splicing in mouse cells. Nature Biotechnology, 2003, 21, 1499-1504.	17.5	24
14	A new dosage test for subtelomeric 4;10 translocations improves conventional diagnosis of facioscapulohumeral muscular dystrophy (FSHD). Journal of Medical Genetics, 1999, 36, 823-8.	3.2	22
15	A New Î²-Thalassemia Mutation Produced by a Single Nucleotide Substitution in the Conserved Dinucleotide Sequence of the IVS-I Consensus Acceptor Site (Agâ†'AA). Hemoglobin, 1990, 14, 431-440.	0.8	16
16	Deep phenotyping of facioscapulohumeral muscular dystrophy type 2 by magnetic resonance imaging. European Journal of Neurology, 2020, 27, 2604-2615.	3.3	16
17	Chromosome 4q35 haplotypes and DNA rearrangements segregating in affected subjects of 19 Italian families with facioscapulohumeral musculatur dystrophy (FSHD). Human Genetics, 1994, 94, 367-374.	3.8	12
18	Frequency and molecular types of deletional Î±-thalassemia in Egypt. Human Genetics, 1989, 81, 211-213.	3.8	9

#	Article	IF	Citations
19	Analysis of Î²-thalassemia mutations in the United Arab Emirates provides evidence for recurrent origin of the IVSINT 5 (G-C) mutation. Human Mutation, 1995, 5, 327-328.	2.5	9
20	Digenic Inheritance of Shortened Repeat Units of the D4Z4 Region and a Loss-of-Function Variant in SMCHD1 in a Family With FSHD. Frontiers in Neurology, 2018, 9, 1027.	2.4	8
21	A new Î²-thalassaemia frameshift mutation detected by PCR after selective hybridization to immobilized oligonucleotides. British Journal of Haematology, 1991, 79, 90-92.	2.5	7
22	ARCHAEAâ€ExPRESs targeting of Î±â€tubulin 4 mRNA: a model for highâ€specificity transâ€splicing. FASEB Journal, 2010, 24, 2976-2984.	0.5	3
23	Highly efficient, in vivo optimized, archaeal endonuclease for controlled RNA splicing in mammalian cells. FASEB Journal, 2013, 27, 3466-3477.	0.5	2