David Valle

List of Publications by Year
in descending order

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79	7,145	117625	76900
papers	citations	34 h-index	74 g-index

82	82	82	12522
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
2	Variantâ€level matching for diagnosis and discovery: Challenges and opportunities. Human Mutation, 2022, , .	2.5	11
3	The impact of GeneMatcher on international data sharing and collaboration. Human Mutation, 2022, , .	2.5	7
4	A promoter variant in the OTC gene associated with late and variable age of onset hyperammonemia. Journal of Inherited Metabolic Disease, 2022, 45, 710-718.	3.6	5
5	IFIH1 loss-of-function variants contribute to very early-onset inflammatory bowel disease. Human Genetics, 2021, 140, 1299-1312.	3.8	17
6	Victor McKusick and his short course. American Journal of Medical Genetics, Part A, 2021, 185, 3242-3252.	1.2	1
7	PhenoDB, GeneMatcher and VariantMatcher, tools for analysis and sharing of sequence data. Orphanet Journal of Rare Diseases, 2021, 16, 365.	2.7	24
8	Response to Biesecker etÂal.. American Journal of Human Genetics, 2021, 108, 1807-1808.	6.2	3
9	The utility of exome sequencing for fetal pleural effusions. Prenatal Diagnosis, 2020, 40, 590-595.	2.3	9
10	The Deep Genome Project. Genome Biology, 2020, 21, 18.	8.8	30
11	Genetic diagnosis in first or second trimester pregnancy loss using exome sequencing: a systematic review of human essential genes. Journal of Assisted Reproduction and Genetics, 2019, 36, 1539-1548.	2.5	25
12	Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS. American Journal of Human Genetics, 2019, 105, 151-165.	6.2	170
13	2018 Victor A. McKusick Leadership Award Introduction: James R. Lupski. American Journal of Human Genetics, 2019, 104, 389-390.	6.2	0
14	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
15	TBX6-associated congenital scoliosis (TACS) as a clinically distinguishable subtype of congenital scoliosis: further evidence supporting the compound inheritance and TBX6 gene dosage model. Genetics in Medicine, 2019, 21, 1548-1558.	2.4	60
16	Familial monophasic acute transverse myelitis due to the pathogenic variant in VPS37A. Neurology: Genetics, 2018, 4, e213.	1.9	4
17	Defect in phosphoinositide signalling through a homozygous variant inPLCB3causes a new form of spondylometaphyseal dysplasia with corneal dystrophy. Journal of Medical Genetics, 2018, 55, 122-130.	3.2	9
18	Identification of STAC3 variants in nonâ€Native American families with overlapping features of Careyâ€“Finemanâ€“Ziter syndrome and Moebius syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2763-2771.	1.2	28

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19	Thorase variants are associated with defects in glutamatergic neurotransmission that can be rescued by Perampanel. Science Translational Medicine, 2017, 9, .	12.4	20
20	Patients with a Kabuki syndrome phenotype demonstrate DNA methylation abnormalities. European Journal of Human Genetics, 2017, 25, 1335-1344.	2.8	52
21	LMNA Sequences of 60,706 Unrelated Individuals Reveal 132 Novel Missense Variants in A-Type Lamins and Suggest a Link between Variant p.G602S and Type 2 Diabetes. Frontiers in Genetics, 2017, 8, 79.	2.3	17
22	A Syndromic Intellectual Disability Disorder Caused by Variants in TELO2 , a Gene Encoding a Component of the TTT Complex. American Journal of Human Genetics, 2016, 98, 909-918.	6.2	35
23	Gonadal mosaicism in ARID1B gene causes intellectual disability and dysmorphic features in three siblings. American Journal of Medical Genetics, Part A, 2016, 170, 156-161.	1.2	16
24	Disorders of Ornithine and Proline Metabolism. , 2016, , 321-331.		4
25	Lessons learned from the search for genes responsible for rare Mendelian disorders. Molecular Genetics & Genomic Medicine, 2016, 4, 371-375.	1.2	7
26	Neuregulin 3 Knockout Mice Exhibit Behaviors Consistent with Psychotic Disorders. Molecular Neuropsychiatry, 2016, 2, 79-87.	2.9	27
27	The management of pregnancy and delivery in 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. , 2016, 170, 1600-1602.		7
28	GeneMatcher Aids in the Identification of a New Malformation Syndrome with Intellectual Disability, Unique Facial Dysmorphisms, and Skeletal and Connective Tissue Abnormalities Caused by De Novo Variants inHNRNPK. Human Mutation, 2015, 36, 1009-1014.	2.5	56
29	GeneMatcher: A Matching Tool for Connecting Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 928-930.	2.5	1,153
30	Identification and Functional Studies of Regulatory Variants Responsible for the Association of <i>NRG3</i> with a Delusion Phenotype in Schizophrenia. Molecular Neuropsychiatry, 2015, 1, 36-46.	2.9	14
31	New Arab family with cerebral dysgenesis, neuropathy, ichthyosis and keratoderma syndrome suggests a possible founder effect for the c.223delG mutation. Journal of Dermatology, 2015, 42, 821-822.	1.2	8
32	Functional Variants in DPYSL2 Sequence Increase Risk of Schizophrenia and Suggest a Link to mTOR Signaling. G3: Genes, Genomes, Genetics, 2015, 5, 61-72.	1.8	39
33	Assessment of incidental findings in 232 whole-exome sequences from the Baylorâ€“Hopkins Center for Mendelian Genomics. Genetics in Medicine, 2015, 17, 782-788.	2.4	41
34	New Tools for Mendelian Disease Gene Identification: PhenoDB Variant Analysis Module; and GeneMatcher, a Web-Based Tool for Linking Investigators with an Interest in the Same Gene. Human Mutation, 2015, 36, 425-431.	2.5	141
35	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	6.2	574
36	2014 Victor A. McKusick Leadership Award1. American Journal of Human Genetics, 2015, 96, 374-376.	6.2	0

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37	A novel bile acid biosynthesis defect due to a deficiency of peroxisomal ABCD3. Human Molecular Genetics, 2015, 24, 361-370.	2.9	115
38	Reduced PLP2 expression increases ER-stress-induced neuronal apoptosis and risk for adverse neurological outcomes after hypoxia ischemia injury. Human Molecular Genetics, 2015, 24, 7221-7226.	2.9	16
39	Enhanced conversion of induced neuronal cells (iN cells) from human fibroblasts: Utility in uncovering cellular deficits in mental illness-associated chromosomal abnormalities. Neuroscience Research, 2015, 101, 57-61.	1.9	14
40	Novel Deletion of <i>SERPINF1</i> Causes Autosomal Recessive Osteogenesis Imperfecta Type VI in Two Brazilian Families. Molecular Syndromology, 2014, 5, 268-275.	0.8	18
41	Mutations in AlstrÃ¶m protein impair terminal differentiation of cardiomyocytes. Nature Communications, 2014, 5, 3416.	12.8	66
42	Mutations in PCYT1A, Encoding a Key Regulator of Phosphatidylcholine Metabolism, Cause Spondylometaphyseal Dysplasia with Cone-Rod Dystrophy. American Journal of Human Genetics, 2014, 94, 105-112.	6.2	53
43	<scp>P</scp> heno <scp>DB</scp> : A New Webâ€Based Tool for the Collection, Storage, and Analysis of Phenotypic Features. Human Mutation, 2013, 34, 566-571.	2.5	64
44	Linkage and association on 8p21.2-p21.1 in schizophrenia. , 2011, 156, 188-197.		26
45	PHR1 is a vesicleâ€bound protein abundantly expressed in mature olfactory neurons. Laryngoscope, 2010, 120, 1002-1010.	2.0	1
46	Familiality of Novel Factorial Dimensions of Schizophrenia. Archives of General Psychiatry, 2009, 66, 591.	12.3	71
47	Fine Mapping on Chromosome 10q22-q23 Implicates Neuregulin 3 in Schizophrenia. American Journal of Human Genetics, 2009, 84, 21-34.	6.2	81
48	Functional genomics and SNP analysis of human genes encoding proline metabolic enzymes. Amino Acids, 2008, 35, 655-664.	2.7	46
49	PRODH variants and risk for schizophrenia. Amino Acids, 2008, 35, 673-679.	2.7	54
50	X chromosome cDNA microarray screening identifies a functional PLP2 promoter polymorphism enriched in patients with X-linked mental retardation. Genome Research, 2007, 17, 641-648.	5.5	16
51	Recurrent 10q22-q23 Deletions: A Genomic Disorder on 10q Associated with Cognitive and Behavioral Abnormalities. American Journal of Human Genetics, 2007, 80, 938-947.	6.2	101
52	Overexpression of proline oxidase induces proline-dependent and mitochondria-mediated apoptosis. Molecular and Cellular Biochemistry, 2007, 295, 85-92.	3.1	98
53	A microdeletion in Xp11.3 accounts for co-segregation of retinitis pigmentosa and mental retardation in a large kindred. American Journal of Medical Genetics, Part A, 2006, 140A, 349-357.	1.2	23
54	Functional Consequences of PRODH Missense Mutations. American Journal of Human Genetics, 2005, 76, 409-420.	6.2	134

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55	Bipolar I Disorder and Schizophrenia: A 440â€“Single-Nucleotide Polymorphism Screen of 64 Candidate Genes among Ashkenazi Jewish Case-Parent Trios. American Journal of Human Genetics, 2005, 77, 918-936.	6.2	358
56	A SCIENCE OF THE INDIVIDUAL: Implications for a Medical School Curriculum. Annual Review of Genomics and Human Genetics, 2005, 6, 313-330.	6.2	34
57	PHR1, a PH Domain-Containing Protein Expressed in Primary Sensory Neurons. Molecular and Cellular Biology, 2004, 24, 9137-9151.	2.3	16
58	Genetics, Individuality, and Medicine in the 21st Century**Previously presented at the annual meeting of The American Society of Human Genetics, in Los Angeles, on November 5, 2003.. American Journal of Human Genetics, 2004, 74, 374-381.	6.2	20
59	Genomewide Linkage Scan for Schizophrenia Susceptibility Loci among Ashkenazi Jewish Families Shows Evidence of Linkage on Chromosome 10q22. American Journal of Human Genetics, 2003, 73, 601-611.	6.2	99
60	Introductory Speech for Charles Scriver**Amended from the introduction previously presented at the annual meeting of The American Society of Human Genetics, in San Diego, on October 16, 2001.. American Journal of Human Genetics, 2002, 70, 314-316.	6.2	0
61	Online Mendelian Inheritance In Man (OMIM). Human Mutation, 2000, 15, 57-61.	2.5	503
62	Genetics, Biology and Disease. Annual Review of Genomics and Human Genetics, 2000, 1, 1-19.	6.2	29
63	PHR1 Encodes an Abundant, Pleckstrin Homology Domain-containing Integral Membrane Protein in the Photoreceptor Outer Segments. Journal of Biological Chemistry, 1999, 274, 35676-35685.	3.4	18
64	40 years of the annual â€˜Bar Harbor Courseâ€™ (1960-1999): a pictorial history. Clinical Genetics, 1999, 55, 398-415.	2.0	6
65	You give me fever. Nature Genetics, 1999, 22, 121-122.	21.4	12
66	Hyperornithinaemia-Âhyperammonaemia-Âhomocitrullinuria syndrome is caused by mutations in a gene encoding a mitochondrial ornithine transporter. Nature Genetics, 1999, 22, 151-158.	21.4	184
67	Gyrate Atrophy of the Choroid and Retina: Lymphocyte Ornithine-Î´-Aminotransferase Activity in Different Mutations and Carriers. Pediatric Research, 1998, 44, 381-385.	2.3	18
68	Isolation of the human PEX12 gene, mutated in group 3 of the peroxisome biogenesis disorders. Nature Genetics, 1997, 15, 385-388.	21.4	142
69	Human PEX7 encodes the peroxisomal PTS2 receptor and is responsible for rhizomelic chondrodysplasia punctata. Nature Genetics, 1997, 15, 369-376.	21.4	415
70	Mutations in PEX1 are the most common cause of peroxisome biogenesis disorders. Nature Genetics, 1997, 17, 445-448.	21.4	233
71	A ubiquitin C-terminal hydrolase gene on the proximal short arm of the X chromosome: implications for X-linked retinal disorders. Human Molecular Genetics, 1996, 5, 533-538.	2.9	31
72	Mutations in the PTS1 receptor gene, PXR1, define complementation group 2 of the peroxisome biogenesis disorders. Nature Genetics, 1995, 9, 115-125.	21.4	432

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73	Mice lacking ornithineâ€“Îâ€“aminoâ€“transferase have paradoxical neonatal hypoornithinaemia and retinal degeneration. Nature Genetics, 1995, 11, 185-190.	21.4	106
74	Transcriptional regulation of the gene for the second component of human complement: Promoter analysis. European Journal of Immunology, 1994, 24, 393-400.	2.9	5
75	Penetrating the peroxisome. Nature, 1993, 361, 682-683.	27.8	73
76	Mutations in the 70K peroxisomal membrane protein gene in Zellweger syndrome. Nature Genetics, 1992, 1, 16-23.	21.4	238
77	Chronic cardiomyopathy and weakness or acute coma in children with a defect in carnitine uptake. Annals of Neurology, 1991, 30, 709-716.	5.3	206
78	Allopurinol-Induced Orotidinuria. New England Journal of Medicine, 1990, 322, 1641-1645.	27.0	124
79	Proline oxidase in cultured mammalian cells. Journal of Cellular Physiology, 1977, 91, 369-376.	4.1	25