

# Silvia Russo

## List of Publications by Year in descending order

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105  
papers

4,436  
citations

156536

32  
h-index

134545

62  
g-index

106  
all docs

106  
docs citations

106  
times ranked

5954  
citing authors

#	ARTICLE	IF	CITATIONS
1	Genes for RNA-binding proteins involved in neural-specific functions and diseases are downregulated in Rubinstein-Taybi iNeurons. <i>Neural Regeneration Research</i> , 2022, 17, 5.	1.6	5
2	Germline variants in genes of the subcortical maternal complex and Multilocus Imprinting Disturbance are associated with miscarriage/infertility or Beckwith-Wiedemann progeny. <i>Clinical Epigenetics</i> , 2022, 14, 43.	1.8	6
3	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. <i>Clinical Epigenetics</i> , 2022, 14, 41.	1.8	14
4	SCN2A Pathogenic Variants and Epilepsy: Heterogeneous Clinical, Genetic and Diagnostic Features. <i>Brain Sciences</i> , 2022, 12, 18.	1.1	5
5	Expanding the Molecular Spectrum of ANKRD11 Gene Defects in 33 Patients with a Clinical Presentation of KBG Syndrome. <i>International Journal of Molecular Sciences</i> , 2022, 23, 5912.	1.8	6
6	Clinical and molecular characterization of patients affected by Beckwith-Wiedemann spectrum conceived through assisted reproduction techniques. <i>Clinical Genetics</i> , 2022, 102, 314-323.	1.0	7
7	Maternal Uniparental Disomy of Chromosome 20 (UPD(20)mat) as Differential Diagnosis of Silver Russell Syndrome: Identification of Three New Cases. <i>Genes</i> , 2021, 12, 588.	1.0	6
8	Histone Deacetylase Inhibitors Ameliorate Morphological Defects and Hypoexcitability of iPSC-Neurons from Rubinstein-Taybi Patients. <i>International Journal of Molecular Sciences</i> , 2021, 22, 5777.	1.8	7
9	Editorial: Overlapping Phenotypes and Genetic Heterogeneity of Rare Neurodevelopmental Disorders. <i>Frontiers in Neurology</i> , 2021, 12, 711288.	1.1	2
10	Interconnected Gene Networks Underpin the Clinical Overlap of HNRNPH1-Related and Rubinstein-Taybi Intellectual Disability Syndromes. <i>Frontiers in Neuroscience</i> , 2021, 15, 745684.	1.4	0
11	Guided Growth in Leg Length Discrepancy in Beckwith-Wiedemann Syndrome: A Consecutive Case Series. <i>Children</i> , 2021, 8, 1152.	0.6	1
12	A novel RAD21 mutation in a boy with mild Cornelia de Lange presentation: Further delineation of the phenotype. <i>European Journal of Medical Genetics</i> , 2020, 63, 103620.	0.7	6
13	Cognitive Profiles and Brain Volume Are Affected in Patients with Silver-Russell Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e1478-e1488.	1.8	4
14	Recombinant Chromosome 7 Driven by Maternal Chromosome 7 Pericentric Inversion in a Girl with Features of Silver-Russell Syndrome. <i>International Journal of Molecular Sciences</i> , 2020, 21, 8487.	1.8	1
15	Sleep disordered breathing and daytime hypoventilation in a male with MECP2 mutation. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2982-2987.	0.7	3
16	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. <i>Clinical Epigenetics</i> , 2020, 12, 139.	1.8	40
17	Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. <i>Molecular Neurobiology</i> , 2020, 57, 3685-3701.	1.9	15
18	DNA Methylation in the Diagnosis of Monogenic Diseases. <i>Genes</i> , 2020, 11, 355.	1.0	28

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19	Comparison of Quantitative Analysis of Methylated Alleles Real-Time PCR and Methylation-Specific MLPA for Molecular Diagnosis of Beckwith-Wiedemann Syndrome. <i>Pathobiology</i> , 2019, 86, 217-224.	1.9	1
20	Pathogenic Variants in STXBP1 and in Genes for GABA <sub>A</sub> Receptor Subunits Cause Atypical Rett/Rett-like Phenotypes. <i>International Journal of Molecular Sciences</i> , 2019, 20, 3621.	1.8	29
21	Generation of three iPSC lines (IAli002, IAli004, IAli003) from Rubinstein-Taybi syndrome 1 patients carrying CREBBP non sense c.4435G>T, p.(Gly1479*) and c.3474G>A, p.(Trp1158*) and missense c.4627G>T, p.(Asp1543Tyr) mutations. <i>Stem Cell Research</i> , 2019, 40, 101553.	0.3	6
22	Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. <i>Genetical Research</i> , 2019, 101, e3.	0.3	17
23	Analysis of the Phenotypes in the Rett Networked Database. <i>International Journal of Genomics</i> , 2019, 2019, 1-9.	0.8	23
24	Molecular Etiology Disclosed by Array CGH in Patients With Silver-Russell Syndrome or Similar Phenotypes. <i>Frontiers in Genetics</i> , 2019, 10, 955.	1.1	11
25	A KHDC3L mutation resulting in recurrent hydatidiform mole causes genome-wide DNA methylation loss in oocytes and persistent imprinting defects post-fertilisation. <i>Genome Medicine</i> , 2019, 11, 84.	3.6	45
26	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. <i>Clinical Epigenetics</i> , 2019, 11, 190.	1.8	22
27	Clinical and molecular diagnosis, screening and management of Beckwith-Wiedemann syndrome: an international consensus statement. <i>Nature Reviews Endocrinology</i> , 2018, 14, 229-249.	4.3	388
28	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. <i>Epigenetics</i> , 2018, 13, 117-121.	1.3	70
29	Segmental Maternal UPD of Chromosome 7q in a Patient With Pendred and Silver Russell Syndromes-Like Features. <i>Frontiers in Genetics</i> , 2018, 9, 600.	1.1	4
30	Workload measurement for molecular genetics laboratory: A survey study. <i>PLoS ONE</i> , 2018, 13, e0206855.	1.1	6
31	Recurrence and Familial Inheritance of Intronic NIPBL Pathogenic Variant Associated With Mild CdLS. <i>Frontiers in Neurology</i> , 2018, 9, 967.	1.1	6
32	Use of nutritional devices in Cornelia de Lange syndrome: Data from a large Italian cohort. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1865-1871.	0.7	2
33	The phenotype of <i>SCN8A</i> developmental and epileptic encephalopathy. <i>Neurology</i> , 2018, 91, e1112-e1124.	1.5	114
34	Generation of the Rubinstein-Taybi syndrome type 2 patient-derived induced pluripotent stem cell line (IAli001-A) carrying the EP300 exon 23 stop mutation c.3829A>T, p.(Lys1277*). <i>Stem Cell Research</i> , 2018, 30, 175-179.	0.3	4
35	Taurine Administration Recovers Motor and Learning Deficits in an Angelman Syndrome Mouse Model. <i>International Journal of Molecular Sciences</i> , 2018, 19, 1088.	1.8	14
36	iPSC-derived neurons of CREBBP - and EP300 -mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. <i>Stem Cell Research</i> , 2018, 30, 130-140.	0.3	19

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37	Clinical Manifestations and Metabolic Outcomes of Seven Adults with Silver-Russell Syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 2225-2233.	1.8	10
38	Nomenclature and definition in asymmetric regional body overgrowth. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1735-1738.	0.7	36
39	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2108-2125.	0.7	69
40	Diagnosis and management of Silver-Russell syndrome: first international consensus statement. <i>Nature Reviews Endocrinology</i> , 2017, 13, 105-124.	4.3	336
41	Cancer Risk in Beckwith-Wiedemann Syndrome: A Systematic Review and Meta-Analysis Outlining a Novel (Epi)Genotype Specific Histotype Targeted Screening Protocol. <i>Journal of Pediatrics</i> , 2016, 176, 142-149.e1.	0.9	119
42	A new prognostic index of severity of intellectual disabilities in Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 179-189.	0.7	7
43	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silver-Russell and Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1377-1387.	1.4	68
44	Thrombocytopenia and Cornelia de Lange syndrome: Still an enigma?. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 130-134.	0.7	4
45	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silver-Russell and Beckwith-Wiedemann syndromes. <i>Clinical Epigenetics</i> , 2016, 8, 23.	1.8	54
46	Sleep disorders in Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 214-221.	0.7	14
47	Functional characterization of CDK5 and CDK5R1 mutations identified in patients with non-syndromic intellectual disability. <i>Journal of Human Genetics</i> , 2016, 61, 283-293.	1.1	12
48	Recommendations of the Scientific Committee of the Italian Beckwith-Wiedemann Syndrome Association on the diagnosis, management and follow-up of the syndrome. <i>European Journal of Medical Genetics</i> , 2016, 59, 52-64.	0.7	76
49	Prenatal molecular testing for Beckwith-Wiedemann and Silver-Russell syndromes: a challenge for molecular analysis and genetic counseling. <i>European Journal of Human Genetics</i> , 2016, 24, 784-793.	1.4	44
50	Blood oxidative stress and metallothionein expression in Rett syndrome: Probing for markers. <i>World Journal of Biological Psychiatry</i> , 2016, 17, 198-209.	1.3	11
51	(Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 183-190.	1.4	113
52	Constitutional de novo deletion of the <i>FBXW7</i> gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. <i>Scientific Reports</i> , 2015, 5, 15454.	1.6	10
53	Antiepileptic drugs in Rett Syndrome. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 446-452.	0.7	13
54	Familial intragenic duplication of <i>ANKRD11</i> underlying three patients of KBG syndrome. <i>Molecular Cytogenetics</i> , 2015, 8, 20.	0.4	18

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55	Epilepsy in Rett syndrome—Lessons from the Rett networked database. <i>Epilepsia</i> , 2015, 56, 569-576.	2.6	47
56	Complex <i>de novo</i> chromosomal rearrangement at 15q11–q13 involving an intrachromosomal triplication in a patient with a severe neuropsychological phenotype: Clinical report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 221-230.	0.7	14
57	Somatic Mosaicism as Modulator of the Global and Intellectual Phenotype in Epimutated Angelman Syndrome Patients. <i>Journal of Intellectual Disability - Diagnosis and Treatment</i> , 2015, 3, 126-137.	0.1	1
58	Overall and allele-specific expression of the <i>SMC1A</i> gene in female Cornelia de Lange syndrome patients and healthy controls. <i>Epigenetics</i> , 2014, 9, 973-979.	1.3	10
59	Cervical spine malformation in cornelia de lange syndrome: A report of three patients. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1520-1524.	0.7	6
60	Think About It. <i>Journal of Child Neurology</i> , 2014, 29, NP74-NP77.	0.7	10
61	Recent insights into genotype–phenotype relationships in patients with Rett syndrome using a fine grain scale. <i>Research in Developmental Disabilities</i> , 2014, 35, 2976-2986.	1.2	40
62	Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. <i>BMC Medical Genetics</i> , 2013, 14, 41.	2.1	15
63	Prevalence of Beckwith–Wiedemann syndrome in North West of Italy. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2481-2486.	0.7	93
64	Cornelia de Lange individuals with new and recurrent <i>SMC1A</i> mutations enhance delineation of mutation repertoire and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2909-2919.	0.7	31
65	Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. <i>European Journal of Medical Genetics</i> , 2013, 56, 138-143.	0.7	24
66	Germline mosaicism in cornelia de lange syndrome: Dilemmas and risk figures. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1825-1826.	0.7	2
67	Quantitative DNA methylation analysis improves epigenotype-phenotype correlations in Beckwith-Wiedemann syndrome. <i>Epigenetics</i> , 2013, 8, 1053-1060.	1.3	33
68	Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. <i>European Journal of Human Genetics</i> , 2012, 20, 734-741.	1.4	23
69	The <i>KCNQ1OT1</i> imprinting control region and non-coding RNA: new properties derived from the study of Beckwith–Wiedemann syndrome and Silver–Russell syndrome cases. <i>Human Molecular Genetics</i> , 2012, 21, 10-25.	1.4	135
70	Deletion of the <i>AP1S2</i> gene in a child with psychomotor delay and hypotonia. <i>European Journal of Medical Genetics</i> , 2012, 55, 124-127.	0.7	8
71	Complex rearrangement involving 9p deletion and duplication in a syndromic patient: Genotype/phenotype correlation and review of the literature. <i>Gene</i> , 2012, 502, 40-45.	1.0	16
72	Juxtaposition of heterochromatic and euchromatic regions by chromosomal translocation mediates a heterochromatic long-range position effect associated with a severe neurological phenotype. <i>Molecular Cytogenetics</i> , 2012, 5, 16.	0.4	22

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73	Rett networked database: An integrated clinical and genetic network of rett syndrome databases. <i>Human Mutation</i> , 2012, 33, 1031-1036.	1.1	19
74	Electroclinical pattern in <i>MECP2</i> duplication syndrome: Eight new reported cases and review of literature. <i>Epilepsia</i> , 2012, 53, 1146-1155.	2.6	34
75	Medical care of adolescents and women with Rett syndrome: An Italian study. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 13-18.	0.7	26
76	Nephrological findings and genotype-phenotype correlation in Beckwith-Wiedemann syndrome. <i>Pediatric Nephrology</i> , 2012, 27, 397-406.	0.9	55
77	Role of UBE3A and ATP10A genes in autism susceptibility region 15q11-q13 in an Italian population: A positive replication for UBE3A. <i>Psychiatry Research</i> , 2011, 185, 33-38.	1.7	29
78	Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. <i>American Journal of Human Genetics</i> , 2010, 86, 185-195.	2.6	220
79	Hypertrophic cardiomyopathy in a girl with Cornelia de Lange syndrome due to mutation in <i>SMC1A</i> . <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2127-2129.	0.7	23
80	Noonan syndrome associated with both a new inactivating familial <i>SOS1</i> and a de novo <i>RAF1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2176-2184.	0.7	20
81	Efficacy of levetiracetam in the treatment of drug-resistant Rett syndrome. <i>Epilepsy Research</i> , 2010, 88, 112-117.	0.8	18
82	Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. <i>BMC Medical Genetics</i> , 2010, 11, 146.	2.1	86
83	Epilepsy in Rett syndrome: Clinical and genetic features. <i>Epilepsy and Behavior</i> , 2010, 19, 296-300.	0.9	68
84	Inherited and Sporadic Epimutations at the <i>IGF2-H19</i> Locus in Beckwith-Wiedemann Syndrome and Wilms' Tumor. <i>Endocrine Development</i> , 2009, 14, 1-9.	1.3	48
85	A <i>CDKL5</i> mutated child with precocious puberty. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1046-1051.	0.7	21
86	Novel mutations in the CDKL5 gene, predicted effects and associated phenotypes. <i>Neurogenetics</i> , 2009, 10, 241-250.	0.7	62
87	Premature chromatid separation is not a useful diagnostic marker for Cornelia de Lange syndrome. <i>Chromosome Research</i> , 2009, 17, 763-771.	1.0	49
88	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwith-Wiedemann syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 611-619.	1.4	194
89	Clinical and electroencephalographic features in patients with CDKL5 mutations: Two new Italian cases and review of the literature. <i>Epilepsy and Behavior</i> , 2008, 12, 326-331.	0.9	36
90	Mechanisms causing imprinting defects in familial Beckwith-Wiedemann syndrome with Wilms' tumour. <i>Human Molecular Genetics</i> , 2007, 16, 254-264.	1.4	100

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91	Prenatal/neonatal pathology in two cases of Cornelia de Lange syndrome harboring novel mutations of NIPBL. <i>Genetics in Medicine</i> , 2007, 9, 188-194.	1.1	18
92	Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. <i>Neurogenetics</i> , 2007, 8, 169-178.	0.7	81
93	X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. <i>Nature Genetics</i> , 2006, 38, 528-530.	9.4	393
94	Mutations and novel polymorphisms in coding regions and UTRs of CDK5R1 and OMG genes in patients with non-syndromic mental retardation. <i>Neurogenetics</i> , 2006, 7, 59-66.	0.7	32
95	A fluorescent method for detecting low-grade 11patLUPD mosaicism in Beckwithâ€™Wiedemann syndrome. <i>Molecular and Cellular Probes</i> , 2003, 17, 295-299.	0.9	11
96	Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. <i>Human Molecular Genetics</i> , 2003, 12, 849-858.	1.4	131
97	Small familial supernumerary ring chromosome 2: FISH characterization and genotype-phenotype correlation. <i>American Journal of Medical Genetics Part A</i> , 2002, 111, 319-323.	2.4	13
98	Refinement of the NHS locus on chromosome Xp22.13 and analysis of five candidate genes. <i>European Journal of Human Genetics</i> , 2002, 10, 516-520.	1.4	23
99	Refined FISH characterization of a de novo 1p22-p36.2 paracentric inversion and associated 1p21-22 deletion in a patient with signs of 1p36 microdeletion syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 99, 308-313.	2.4	7
100	Molecular characterization of FRAXE-positive subjects with mental impairment in two unrelated Italian families. <i>American Journal of Medical Genetics Part A</i> , 1998, 75, 304-308.	2.4	13
101	Search for mutations in pancreatic sufficient cystic fibrosis Italian patients: detection of 90% of molecular defects and identification of three novel mutations. <i>Human Genetics</i> , 1995, 96, 312-8.	1.8	23
102	Genetic diagnosis by chorionic villus sampling before 8 gestational weeks: Efficiency, reliability, and risks on 317 completed pregnancies. <i>Prenatal Diagnosis</i> , 1992, 12, 789-799.	1.1	86
103	Frequency of Cystic Fibrosis Mutations Among Italian Patients. <i>Advances in Experimental Medicine and Biology</i> , 1991, 290, 387-390.	0.8	0
104	Frequency of the $\Delta$ F508 mutation in a sample of 175 Italian cystic fibrosis patients. <i>Human Genetics</i> , 1990, 85, 400-402.	1.8	9
105	Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. <i>Human Genetics</i> , 1990, 85, 275-8.	1.8	3