# Silvia Russo

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5216342/publications.pdf

Version: 2024-02-01

| 105 | 4,436 | 136940 | 118840 |
|---|---|---|---|
| papers | citations | 32 h-index | 62 g-index |

| 106 | 106 | 106 | 5536 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Genes for RNA-binding proteins involved in neural-specific functions and diseases are downregulated in Rubinstein-Taybi iNeurons. Neural Regeneration Research, 2022, 17, 5. | 3.0 | 5 |
| 2 | Germline variants in genes of the subcortical maternal complex and Multilocus Imprinting Disturbance are associated with miscarriage/infertility or Beckwithâ€“Wiedemann progeny. Clinical Epigenetics, 2022, 14, 43. | 4.1 | 6 |
| 3 | Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41. | 4.1 | 14 |
| 4 | SCN2A Pathogenic Variants and Epilepsy: Heterogeneous Clinical, Genetic and Diagnostic Features. Brain Sciences, 2022, 12, 18. | 2.3 | 5 |
| 5 | Expanding the Molecular Spectrum of ANKRD11 Gene Defects in 33 Patients with a Clinical Presentation of KBG Syndrome. International Journal of Molecular Sciences, 2022, 23, 5912. | 4.1 | 6 |
| 6 | Clinical and molecular characterization of patients affected by <scp>Beckwithâ€Wiedemann</scp> spectrum conceived through assisted reproduction techniques. Clinical Genetics, 2022, 102, 314-323. | 2.0 | 7 |
| 7 | Maternal Uniparental Disomy of Chromosome 20 (UPD(20)mat) as Differential Diagnosis of Silver Russell Syndrome: Identification of Three New Cases. Genes, 2021, 12, 588. | 2.4 | 6 |
| 8 | Histone Deacetylase Inhibitors Ameliorate Morphological Defects and Hypoexcitability of iPSC-Neurons from Rubinstein-Taybi Patients. International Journal of Molecular Sciences, 2021, 22, 5777. | 4.1 | 7 |
| 9 | Editorial: Overlapping Phenotypes and Genetic Heterogeneity of Rare Neurodevelopmental Disorders. Frontiers in Neurology, 2021, 12, 711288. | 2.4 | 2 |
| 10 | Interconnected Gene Networks Underpin the Clinical Overlap of HNRNPH1-Related and Rubinsteinâ€“Taybi Intellectual Disability Syndromes. Frontiers in Neuroscience, 2021, 15, 745684. | 2.8 | 0 |
| 11 | Guided Growth in Leg Length Discrepancy in Beckwith-Wiedemann Syndrome: A Consecutive Case Series. Children, 2021, 8, 1152. | 1.5 | 1 |
| 12 | A novel RAD21 mutation in a boy with mild Cornelia de Lange presentation: Further delineation of the phenotype. European Journal of Medical Genetics, 2020, 63, 103620. | 1.3 | 6 |
| 13 | Cognitive Profiles and Brain Volume Are Affected in Patients with Silverâ€“Russell Syndrome. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e1478-e1488. | 3.6 | 4 |
| 14 | Recombinant Chromosome 7 Driven by Maternal Chromosome 7 Pericentric Inversion in a Girl with Features of Silver-Russell Syndrome. International Journal of Molecular Sciences, 2020, 21, 8487. | 4.1 | 1 |
| 15 | Sleep disordered breathing and daytime hypoventilation in a male with MECP2 mutation. American Journal of Medical Genetics, Part A, 2020, 182, 2982-2987. | 1.2 | 3 |
| 16 | Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. Clinical Epigenetics, 2020, 12, 139. | 4.1 | 40 |
| 17 | Transcriptome Analysis of iPSC-Derived Neurons from Rubinstein-Taybi Patients Reveals Deficits in Neuronal Differentiation. Molecular Neurobiology, 2020, 57, 3685-3701. | 4.0 | 15 |
| 18 | DNA Methylation in the Diagnosis of Monogenic Diseases. Genes, 2020, 11, 355. | 2.4 | 28 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Comparison of Quantitative Analysis of Methylated Alleles Real-Time PCR and Methylation-Specific MLPA for Molecular Diagnosis of Beckwith-Wiedemann Syndrome. Pathobiology, 2019, 86, 217-224. | 3.8 | 1 |
| 20 | Pathogenic Variants in STXBP1 and in Genes for GABAa Receptor Subunities Cause Atypical Rett/Rett-like Phenotypes. International Journal of Molecular Sciences, 2019, 20, 3621. | 4.1 | 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. Stem Cell Research, 2019, 40, 101553. | 0.7 | 6 |
| 22 | Discrepant molecular and clinical diagnoses in Beckwith-Wiedemann and Silver-Russell syndromes. Genetical Research, 2019, 101, e3. | 0.9 | 17 |
| 23 | Analysis of the Phenotypes in the Rett Networked Database. International Journal of Genomics, 2019, 2019, 1-9. | 1.6 | 23 |
| 24 | Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955. | 2.3 | 11 |
| 25 | A KHDC3L mutation resulting in recurrent hydatidiform mole causes genome-wide DNA methylation loss in oocytes and persistent imprinting defects post-fertilisation. Genome Medicine, 2019, 11, 84. | 8.2 | 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. Clinical Epigenetics, 2019, 11, 190. | 4.1 | 22 |
| 27 | Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249. | 9.6 | 388 |
| 28 | Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. Epigenetics, 2018, 13, 117-121. | 2.7 | 70 |
| 29 | Segmental Maternal UPD of Chromosome 7q in a Patient With Pendred and Silver Russell Syndromes-Like Features. Frontiers in Genetics, 2018, 9, 600. | 2.3 | 4 |
| 30 | Workload measurement for molecular genetics laboratory: A survey study. PLoS ONE, 2018, 13, e0206855. | 2.5 | 6 |
| 31 | Recurrence and Familial Inheritance of Intronic NIPBL Pathogenic Variant Associated With Mild CdLS. Frontiers in Neurology, 2018, 9, 967. | 2.4 | 6 |
| 32 | Use of nutritional devices in Cornelia de Lange syndrome: Data from a large Italian cohort. American Journal of Medical Genetics, Part A, 2018, 176, 1865-1871. | 1.2 | 2 |
| 33 | The phenotype of SCN8A developmental and epileptic encephalopathy. Neurology, 2018, 91, e1112-e1124. | 1.1 | 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.3829Aâ€>â€T, p.(Lys1277*). Stem Cell Research, 2018, 30, 175-179. | 0.7 | 4 |
| 35 | Taurine Administration Recovers Motor and Learning Deficits in an Angelman Syndrome Mouse Model. International Journal of Molecular Sciences, 2018, 19, 1088. | 4.1 | 14 |
| 36 | iPSC-derived neurons of CREBBP - and EP300 -mutated Rubinstein-Taybi syndrome patients show morphological alterations and hypoexcitability. Stem Cell Research, 2018, 30, 130-140. | 0.7 | 19 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Clinical Manifestations and Metabolic Outcomes of Seven Adults with Silver-Russell Syndrome. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2225-2233. | 3.6 | 10 |
| 38 | Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738. | 1.2 | 36 |
| 39 | Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125. | 1.2 | 69 |
| 40 | Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124. | 9.6 | 336 |
| 41 | Cancer Risk in Beckwith-Wiedemann Syndrome: A Systematic Review and Meta-Analysis Outlining a Novel (Epi)Genotype Specific Histotype Targeted Screening Protocol. Journal of Pediatrics, 2016, 176, 142-149.e1. | 1.8 | 119 |
| 42 | A new prognostic index of severity of intellectual disabilities in Cornelia de Lange syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 179-189. | 1.6 | 7 |
| 43 | EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€“Russell and Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387. | 2.8 | 68 |
| 44 | Thrombocytopenia and Cornelia de Lange syndrome: Still an enigma?. American Journal of Medical Genetics, Part A, 2016, 170, 130-134. | 1.2 | 4 |
| 45 | A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silverâ€“Russell and Beckwithâ€“Wiedemann syndromes. Clinical Epigenetics, 2016, 8, 23. | 4.1 | 54 |
| 46 | Sleep disorders in Cornelia de Lange syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 214-221. | 1.6 | 14 |
| 47 | Functional characterization of CDK5 and CDK5R1 mutations identified in patients with non-syndromic intellectual disability. Journal of Human Genetics, 2016, 61, 283-293. | 2.3 | 12 |
| 48 | Recommendations of the Scientific Committee of the Italian Beckwithâ€“Wiedemann Syndrome Association on the diagnosis, management and follow-up of the syndrome. European Journal of Medical Genetics, 2016, 59, 52-64. | 1.3 | 76 |
| 49 | Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793. | 2.8 | 44 |
| 50 | Blood oxidative stress and metallothionein expression in Rett syndrome: Probing for markers. World Journal of Biological Psychiatry, 2016, 17, 198-209. | 2.6 | 11 |
| 51 | (Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190. | 2.8 | 113 |
| 52 | Constitutional de novo deletion of the FBXW7 gene in a patient with focal segmental glomerulosclerosis and multiple primitive tumors. Scientific Reports, 2015, 5, 15454. | 3.3 | 10 |
| 53 | Antiepileptic drugs in Rett Syndrome. European Journal of Paediatric Neurology, 2015, 19, 446-452. | 1.6 | 13 |
| 54 | Familial intragenic duplication of ANKRD11 underlying three patients of KBG syndrome. Molecular Cytogenetics, 2015, 8, 20. | 0.9 | 18 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Epilepsy in Rett syndromeâ€"Lessons from the Rett networked database. Epilepsia, 2015, 56, 569-576. | 5.1 | 47 |
| 56 | Complex de novo chromosomal rearrangement at 15q11â€"q13 involving an intrachromosomal triplication in a patient with a severe neuropsychological phenotype: Clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2015, 167, 221-230. | 1.2 | 14 |
| 57 | Somatic Mosaicism as Modulator of the Global and Intellectual Phenotype in Epimutated Angelman Syndrome Patients. Journal of Intellectual Disability - Diagnosis and Treatment, 2015, 3, 126-137. | 0.3 | 1 |
| 58 | Overall and allele-specific expression of theSMC1Agene in female Cornelia de Lange syndrome patients and healthy controls. Epigenetics, 2014, 9, 973-979. | 2.7 | 10 |
| 59 | Cervical spine malformation in cornelia de lange syndrome: A report of three patients. American Journal of Medical Genetics, Part A, 2014, 164, 1520-1524. | 1.2 | 6 |
| 60 | Think About It. Journal of Child Neurology, 2014, 29, NP74-NP77. | 1.4 | 10 |
| 61 | Recent insights into genotypeâ€"phenotype relationships in patients with Rett syndrome using a fine grain scale. Research in Developmental Disabilities, 2014, 35, 2976-2986. | 2.2 | 40 |
| 62 | Genomic imbalances in patients with a clinical presentation in the spectrum of Cornelia de Lange syndrome. BMC Medical Genetics, 2013, 14, 41. | 2.1 | 15 |
| 63 | Prevalence of beckwithâ€"wiedemann syndrome in North West of Italy. American Journal of Medical Genetics, Part A, 2013, 161, 2481-2486. | 1.2 | 93 |
| 64 | Cornelia de Lange individuals with new and recurrent SMC1A mutations enhance delineation of mutation repertoire and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2013, 161, 2909-2919. | 1.2 | 31 |
| 65 | Molecular characterization of a mosaic NIPBL deletion in a Cornelia de Lange patient with severe phenotype. European Journal of Medical Genetics, 2013, 56, 138-143. | 1.3 | 24 |
| 66 | Germline mosaicism in cornelia de lange syndrome: Dilemmas and risk figures. American Journal of Medical Genetics, Part A, 2013, 161, 1825-1826. | 1.2 | 2 |
| 67 | Quantitative DNA methylation analysis improves epigenotype-phenotype correlations in Beckwith-Wiedemann syndrome. Epigenetics, 2013, 8, 1053-1060. | 2.7 | 33 |
| 68 | Intragenic and large NIPBL rearrangements revealed by MLPA in Cornelia de Lange patients. European Journal of Human Genetics, 2012, 20, 734-741. | 2.8 | 23 |
| 69 | The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwithâ€"Wiedemann syndrome and Silverâ€"Russell syndrome cases. Human Molecular Genetics, 2012, 21, 10-25. | 2.9 | 135 |
| 70 | Deletion of the AP1S2 gene in a child with psychomotor delay and hypotonia. European Journal of Medical Genetics, 2012, 55, 124-127. | 1.3 | 8 |
| 71 | Complex rearrangement involving 9p deletion and duplication in a syndromic patient: Genotype/phenotype correlation and review of the literature. Gene, 2012, 502, 40-45. | 2.2 | 16 |
| 72 | Juxtaposition of heterochromatic and euchromatic regions by chromosomal translocation mediates a heterochromatic long-range position effect associated with a severe neurological phenotype. Molecular Cytogenetics, 2012, 5, 16. | 0.9 | 22 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Rett networked database: An integrated clinical and genetic network of rett syndrome databases. Human Mutation, 2012, 33, 1031-1036. | 2.5 | 19 |
| 74 | Electroclinical pattern in MECP2 duplication syndrome: Eight new reported cases and review of literature. Epilepsia, 2012, 53, 1146-1155. | 5.1 | 34 |
| 75 | Medical care of adolescents and women with Rett syndrome: An Italian study. American Journal of Medical Genetics, Part A, 2012, 158A, 13-18. | 1.2 | 26 |
| 76 | Nephrological findings and genotypeâ€“phenotype correlation in Beckwithâ€“Wiedemann syndrome. Pediatric Nephrology, 2012, 27, 397-406. | 1.7 | 55 |
| 77 | Role of UBE3A and ATP10A genes in autism susceptibility region 15q11-q13 in an Italian population: A positive replication for UBE3A. Psychiatry Research, 2011, 185, 33-38. | 3.3 | 29 |
| 78 | Mutations in the Small GTPase Gene RAB39B Are Responsible for X-linked Mental Retardation Associated with Autism, Epilepsy, and Macrocephaly. American Journal of Human Genetics, 2010, 86, 185-195. | 6.2 | 220 |
| 79 | Hypertrophic cardiomyopathy in a girl with Cornelia de Lange syndrome due to mutation in SMC1A. American Journal of Medical Genetics, Part A, 2010, 152A, 2127-2129. | 1.2 | 23 |
| 80 | Noonan syndrome associated with both a new Jnkâ€activating familial SOS1 and a de novo RAF1 mutations. American Journal of Medical Genetics, Part A, 2010, 152A, 2176-2184. | 1.2 | 20 |
| 81 | Efficacy of levetiracetam in the treatment of drug-resistant Rett syndrome. Epilepsy Research, 2010, 88, 112-117. | 1.6 | 18 |
| 82 | Genetic investigations on 8 patients affected by ring 20 chromosome syndrome. BMC Medical Genetics, 2010, 11, 146. | 2.1 | 86 |
| 83 | Epilepsy in Rett syndrome: Clinical and genetic features. Epilepsy and Behavior, 2010, 19, 296-300. | 1.7 | 68 |
| 84 | Inherited and Sporadic Epimutations at the IGF2-H19 Locus in Beckwith-Wiedemann Syndrome and Wilms’ Tumor. Endocrine Development, 2009, 14, 1-9. | 1.3 | 48 |
| 85 | A CDKL5 mutated child with precocious puberty. American Journal of Medical Genetics, Part A, 2009, 149A, 1046-1051. | 1.2 | 21 |
| 86 | Novel mutations in the CDKL5 gene, predicted effects and associated phenotypes. Neurogenetics, 2009, 10, 241-250. | 1.4 | 62 |
| 87 | Premature chromatid separation is not a useful diagnostic marker for Cornelia de Lange syndrome. Chromosome Research, 2009, 17, 763-771. | 2.2 | 49 |
| 88 | Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619. | 2.8 | 194 |
| 89 | Clinical and electroencephalographic features in patients with CDKL5 mutations: Two new Italian cases and review of the literature. Epilepsy and Behavior, 2008, 12, 326-331. | 1.7 | 36 |
| 90 | Mechanisms causing imprinting defects in familial Beckwithâ€“Wiedemann syndrome with Wilms' tumour. Human Molecular Genetics, 2007, 16, 254-264. | 2.9 | 100 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Prenatal/neonatal pathology in two cases of Cornelia de Lange syndrome harboring novel mutations of NIPBL. Genetics in Medicine, 2007, 9, 188-194. | 2.4 | 18 |
| 92 | Evaluation of autism traits in Angelman syndrome: a resource to unfold autism genes. Neurogenetics, 2007, 8, 169-178. | 1.4 | 81 |
| 93 | X-linked Cornelia de Lange syndrome owing to SMC1L1 mutations. Nature Genetics, 2006, 38, 528-530. | 21.4 | 393 |
| 94 | Mutations and novel polymorphisms in coding regions and UTRs of CDK5R1 and OMG genes in patients with non-syndromic mental retardation. Neurogenetics, 2006, 7, 59-66. | 1.4 | 32 |
| 95 | A fluorescent method for detecting low-grade 11patUPD mosaicism in Beckwithâ€“Wiedemann syndrome. Molecular and Cellular Probes, 2003, 17, 295-299. | 2.1 | 11 |
| 96 | Genomic inversions of human chromosome 15q11-q13 in mothers of Angelman syndrome patients with class II (BP2/3) deletions. Human Molecular Genetics, 2003, 12, 849-858. | 2.9 | 131 |
| 97 | Small familial supernumerary ring chromosome 2: FISH characterization and genotypeâ€phenotype correlation. American Journal of Medical Genetics Part A, 2002, 111, 319-323. | 2.4 | 13 |
| 98 | Refinement of the NHS locus on chromosome Xp22.13 and analysis of five candidate genes. European Journal of Human Genetics, 2002, 10, 516-520. | 2.8 | 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. American Journal of Medical Genetics Part A, 2001, 99, 308-313. | 2.4 | 7 |
| 100 | Molecular characterization of FRAXE-positive subjects with mental impairment in two unrelated Italian families. American Journal of Medical Genetics Part A, 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. Human Genetics, 1995, 96, 312-8. | 3.8 | 23 |
| 102 | Genetic diagnosis by chorionic villus sampling before 8 gestational weeks: Efficiency, reliability, and risks on 317 completed pregnancies. Prenatal Diagnosis, 1992, 12, 789-799. | 2.3 | 86 |
| 103 | Frequency of Cystic Fibrosis Mutations Among Italian Patients. Advances in Experimental Medicine and Biology, 1991, 290, 387-390. | 1.6 | 0 |
| 104 | Frequency of the Î”F508 mutation in a sample of 175 Italian cystic fibrosis patients. Human Genetics, 1990, 85, 400-402. | 3.8 | 9 |
| 105 | Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. Human Genetics, 1990, 85, 275-8. | 3.8 | 3 |