

Domenica Taruscio

List of Publications by Year in descending order

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150
papers

4,452
citations

101543

36
h-index

144013

57
g-index

155
all docs

155
docs citations

155
times ranked

6878
citing authors

#	ARTICLE	IF	CITATIONS
1	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. <i>American Journal of Human Genetics</i> , 2017, 100, 695-705.	6.2	305
2	Cell Cycle-Dependent Distribution of Telomeres, Centromeres, and Chromosome-Specific Subsatellite Domains in the Interphase Nucleus of Mouse Lymphocytes. <i>Experimental Cell Research</i> , 1993, 205, 142-151.	2.6	159
3	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. <i>Journal of General Internal Medicine</i> , 2014, 29, 780-787.	2.6	159
4	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. <i>Clinical and Translational Science</i> , 2018, 11, 21-27.	3.1	154
5	A Population-Based Study on the Frequency of Additional Congenital Malformations in Infants with Congenital Hypothyroidism: Data from the Italian Registry for Congenital Hypothyroidism (1991-1998). <i>Journal of Clinical Endocrinology and Metabolism</i> , 2002, 87, 557-562.	3.6	128
6	Recommendations for Improving the Quality of Rare Disease Registries. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 1644.	2.6	116
7	The TREAT-NMD Duchenne Muscular Dystrophy Registries: Conception, Design, and Utilization by Industry and Academia. <i>Human Mutation</i> , 2013, 34, 1449-1457.	2.5	94
8	Newborn screening programmes in Europe; arguments and efforts regarding harmonization. Part 2 â€“ From screening laboratory results to treatment, follow-up and quality assurance. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 613-625.	3.6	88
9	The possible role of endocrine disrupting chemicals in the aetiology of cryptorchidism and hypospadias: a population-based case-control study in rural Sicily. <i>Journal of Developmental and Physical Disabilities</i> , 2007, 30, 3-13.	3.6	82
10	Integration site preferences of endogenous retroviruses. <i>Chromosoma</i> , 1991, 101, 141-156.	2.2	76
11	Health-Related Quality of Life in Patients with Neurofibromatosis Type 1. <i>Dermatology</i> , 2009, 218, 215-220.	2.1	73
12	Cryptorchidism and hypospadias in the Sicilian district of Ragusa and the use of pesticides. <i>Reproductive Toxicology</i> , 2006, 22, 8-12.	2.9	70
13	The need for worldwide policy and action plans for rare diseases. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2012, 101, 805-807.	1.5	69
14	Undiagnosed Diseases Network International (UDNI): White paper for global actions to meet patient needs. <i>Molecular Genetics and Metabolism</i> , 2015, 116, 223-225.	1.1	69
15	Identification of Key Regions and Genes Important in the Pathogenesis of SÃ©zary Syndrome by Combining Genomic and Expression Microarrays. <i>Cancer Research</i> , 2009, 69, 8438-8446.	0.9	68
16	Altered microRNA Expression Patterns in Hepatoblastoma Patients. <i>Translational Oncology</i> , 2009, 2, 157-163.	3.7	68
17	Social/economic costs and health-related quality of life in patients with Duchenne muscular dystrophy in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 19-29.	2.8	59
18	Allelotype of pancreatic acinar cell carcinoma. <i>International Journal of Cancer</i> , 2000, 88, 772-777.	5.1	57

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19	Oxidative stress activation of miR-125b is part of the molecular switch for Hailey-Hailey disease manifestation. <i>Experimental Dermatology</i> , 2011, 20, 932-937.	2.9	57
20	Data Quality in Rare Diseases Registries. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 149-164.	1.6	56
21	Human dopamine D5 receptor pseudogenes. <i>Gene</i> , 1991, 109, 211-218.	2.2	53
22	Social/economic costs and quality of life in patients with haemophilia in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 53-65.	2.8	53
23	Genetic and physical map of the interferon region on chromosome 9p. <i>Genomics</i> , 1992, 14, 105-112.	2.9	52
24	Social/economic costs and health-related quality of life in patients with epidermolysis bullosa in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 31-42.	2.8	50
25	In utero exposure to di-(2-ethylhexyl) phthalate affects liver morphology and metabolism in post-natal CD-1 mice. <i>Reproductive Toxicology</i> , 2010, 29, 427-432.	2.9	49
26	A framework to start the debate on neonatal screening policies in the EU: an Expert Opinion Document. <i>European Journal of Human Genetics</i> , 2014, 22, 12-17.	2.8	49
27	Improving the informed consent process in international collaborative rare disease research: effective consent for effective research. <i>European Journal of Human Genetics</i> , 2016, 24, 1248-1254.	2.8	47
28	Malignant histiocytosis (true histiocytic lymphoma) clinicopathological study of 25 cases. <i>Histopathology</i> , 1985, 9, 905-920.	2.9	46
29	The risk of re-identification versus the need to identify individuals in rare disease research. <i>European Journal of Human Genetics</i> , 2016, 24, 1553-1558.	2.8	46
30	Maternal diet and the risk of hypospadias and cryptorchidism in the offspring. <i>Paediatric and Perinatal Epidemiology</i> , 2008, 22, 249-260.	1.7	45
31	Mortality associated with neurofibromatosis type 1: A study based on Italian death certificates (1995-2006). <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 11.	2.7	44
32	Three cases of rare salivary gland tumours: a molecular study of TP53, CDKN2A/ARF, RAS, BRAF, PTEN, MAPK2 and EGFR genes. <i>Oncology Reports</i> , 2011, 26, 3-11.	2.6	42
33	The EPIRARE proposal of a set of indicators and common data elements for the European platform for rare disease registration. <i>Archives of Public Health</i> , 2014, 72, 35.	2.4	41
34	Social/economic costs and health-related quality of life of mucopolysaccharidosis patients and their caregivers in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 89-98.	2.8	41
35	Meeting Patients's Right to the Correct Diagnosis: Ongoing International Initiatives on Undiagnosed Rare Diseases and Ethical and Social Issues. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 2072.	2.6	40
36	European Recommendations for Primary Prevention of Congenital Anomalies: A Joined Effort of EUROCAT and EUROPLAN Projects to Facilitate Inclusion of This Topic in the National Rare Disease Plans. <i>Public Health Genomics</i> , 2014, 17, 115-123.	1.0	39

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37	Factors regulating endogenous retroviral sequences in human and mouse. <i>Cytogenetic and Genome Research</i> , 2004, 105, 351-362.	1.1	38
38	Delphi approach to select rare diseases for a European representative survey. The BURQOL-RD study. <i>Health Policy</i> , 2012, 108, 19-26.	3.0	38
39	Social/economic costs and health-related quality of life in patients with cystic fibrosis in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 7-18.	2.8	38
40	Test Pricing and Reimbursement in Genomic Medicine: Towards a General Strategy. <i>Public Health Genomics</i> , 2016, 19, 352-363.	1.0	37
41	Progress, challenges and global approaches to rare diseases. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2021, 110, 2711-2716.	1.5	37
42	Nonrandom gain of chromosome 7 in central neurocytoma: A chromosomal analysis and fluorescence in situ hybridization study. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 1997, 430, 47-51.	2.8	36
43	Complex multipathways alterations and oxidative stress are associated with Hailey-Hailey disease. <i>British Journal of Dermatology</i> , 2010, 162, 518-526.	1.5	35
44	MicroRNA profiling of multiple osteochondromas: identification of disease-specific and normal cartilage signatures. <i>Clinical Genetics</i> , 2010, 78, 507-516.	2.0	35
45	Social/economic costs and health-related quality of life in patients with fragile X syndrome in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 43-52.	2.8	35
46	Detection of Trisomy 12 on Ovarian Sex Cord Stromal Tumors by Fluorescence In Situ Hybridization. <i>Diagnostic Molecular Pathology</i> , 1993, 2, 94-98.	2.1	34
47	The Current Situation and Needs of Rare Disease Registries in Europe. <i>Public Health Genomics</i> , 2013, 16, 288-298.	1.0	33
48	The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. <i>European Journal of Human Genetics</i> , 2018, 26, 631-643.	2.8	33
49	The case for open science: rare diseases. <i>JAMIA Open</i> , 2020, 3, 472-486.	2.0	33
50	Nonrandom chromosomal imbalances in primary mediastinal b-cell lymphoma detected by arbitrarily primed PCR fingerprinting. , 1999, 26, 203-209.		32
51	Social/economic costs and health-related quality of life in patients with Prader-Willi syndrome in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 99-108.	2.8	31
52	A call for global action for rare diseases in Africa. <i>Nature Genetics</i> , 2020, 52, 21-26.	21.4	31
53	National Registries of Rare Diseases in Europe: An Overview of the Current Situation and Experiences. <i>Public Health Genomics</i> , 2015, 18, 20-25.	1.0	30
54	Quality of life assessment in a sample of patients affected by Prader-Willi syndrome. <i>Journal of Paediatrics and Child Health</i> , 2007, 43, 826-830.	0.8	29

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55	Chromosomal alterations detected by comparative genomic hybridization in nonfunctioning endocrine pancreatic tumors. <i>Cancer Genetics and Cytogenetics</i> , 2005, 156, 23-30.	1.0	28
56	National Plans and Strategies on Rare Diseases in Europe. <i>Advances in Experimental Medicine and Biology</i> , 2010, 686, 475-491.	1.6	28
57	Social/economic costs and health-related quality of life in patients with scleroderma in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 109-117.	2.8	28
58	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. <i>BioMed Research International</i> , 2017, 2017, 1-13.	1.9	28
59	Policies on Conflicts of Interest in Health Care Guideline Development: A Cross-Sectional Analysis. <i>PLoS ONE</i> , 2016, 11, e0166485.	2.5	28
60	Human Endogenous Retroviral Sequences: Possible Roles in Reproductive Physiopathology ¹ . <i>Biology of Reproduction</i> , 1998, 59, 713-724.	2.7	27
61	Pancreatic acinar carcinoma shows a distinct pattern of chromosomal imbalances by comparative genomic hybridization. <i>Genes Chromosomes and Cancer</i> , 2000, 28, 294-299.	2.8	27
62	EQUAL-qual: A European Program for External Quality Assessment of Genomic DNA Extraction and PCR Amplification. <i>Clinical Chemistry</i> , 2007, 53, 1349-1357.	3.2	27
63	Folic acid and primary prevention of birth defects. <i>BioFactors</i> , 2011, 37, 280-284.	5.4	27
64	Numerical chromosomal aberrations in thyroid tumors detected by double fluorescence in situ hybridization. <i>Genes Chromosomes and Cancer</i> , 1994, 9, 180-185.	2.8	26
65	Alteration of chromosome arm 6p is characteristic of primary mediastinal B-cell lymphoma, as identified by genome-wide allelotyping. <i>Genes Chromosomes and Cancer</i> , 2001, 31, 191-195.	2.8	26
66	Neoplastic and reactive follicles within B-cell malignant lymphomas. A morphological and immunological study of 30 cases. <i>Hematological Oncology</i> , 1985, 3, 243-260.	1.7	25
67	Rare diseases and orphan drugs. <i>Annali Dell'Istituto Superiore Di Sanita</i> , 2011, 47, 83-93.	0.4	25
68	Initiating an undiagnosed diseases program in the Western Australian public health system. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 83.	2.7	24
69	The human Per1 gene: genomic organization and promoter analysis of the first human orthologue of the <i>Drosophila</i> period gene. <i>Gene</i> , 2000, 253, 161-170.	2.2	23
70	Quality assessment in cytogenetic and molecular genetic testing: the experience of the Italian Project on Standardisation and Quality Assurance. <i>Clinical Chemistry and Laboratory Medicine</i> , 2004, 42, 915-21.	2.3	23
71	Primary Prevention of Congenital Anomalies: Recommendable, Feasible and Achievable. <i>Public Health Genomics</i> , 2015, 18, 184-191.	1.0	23
72	The social burden and quality of life of patients with haemophilia in Italy. <i>Blood Transfusion</i> , 2014, 12 Suppl 3, s567-75.	0.4	22

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73	Rare Disease Registries Classification and Characterization: A Data Mining Approach. <i>Public Health Genomics</i> , 2015, 18, 113-122.	1.0	21
74	Report of an International Survey of Molecular Genetic Testing Laboratories. <i>Public Health Genomics</i> , 2007, 10, 123-131.	1.0	20
75	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 55-94.	1.6	20
76	Organization and integration sites in the human genome of endogenous retroviral sequences belonging to HERV-E family. <i>Mammalian Genome</i> , 2002, 13, 216-222.	2.2	18
77	The Italian National Rare Diseases Registry. <i>Blood Transfusion</i> , 2014, 12 Suppl 3, s606-13.	0.4	18
78	Undiagnosed Diseases: Italy-US Collaboration and International Efforts to Tackle Rare and Common Diseases Lacking a Diagnosis. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 25-38.	1.6	17
79	Primary Sclerosing Cholangitis: Burden of Disease and Mortality Using Data from the National Rare Diseases Registry in Italy. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 3095.	2.6	17
80	A Model for the European Platform for Rare Disease Registries. <i>Public Health Genomics</i> , 2013, 16, 299-304.	1.0	16
81	Current trends in biobanking for rare diseases: a review. <i>Journal of Biorepository Science for Applied Medicine</i> , 0, , 49.	0.2	16
82	The Quality of Rare Disease Registries: Evaluation and Characterization. <i>Public Health Genomics</i> , 2016, 19, 108-115.	1.0	16
83	Social/economic costs and health-related quality of life in patients with histiocytosis in Europe. <i>European Journal of Health Economics</i> , 2016, 17, 67-78.	2.8	16
84	The Task-force in Europe for Drug Development for the Young (TEDDY) Network of Excellence. <i>Paediatric Drugs</i> , 2009, 11, 18-21.	3.1	15
85	EUROPLAN: A Project to Support the Development of National Plans on Rare Diseases in Europe. <i>Public Health Genomics</i> , 2013, 16, 278-287.	1.0	15
86	Reimbursed Price of Orphan Drugs: Current Strategies and Potential Improvements. <i>Public Health Genomics</i> , 2017, 20, 1-8.	1.0	15
87	The Occurrence of 275 Rare Diseases and 47 Rare Disease Groups in Italy. Results from the National Registry of Rare Diseases. <i>International Journal of Environmental Research and Public Health</i> , 2018, 15, 1470.	2.6	15
88	Improving diagnosis for rare diseases: the experience of the Italian undiagnosed Rare diseases network. <i>Italian Journal of Pediatrics</i> , 2020, 46, 130.	2.6	14
89	Centres of Expertise and European Reference Networks: key issues in the field of rare diseases. The EUCERD Recommendations. <i>Blood Transfusion</i> , 2014, 12 Suppl 3, s621-5.	0.4	14
90	A case report of a patient with microcephaly, facial dysmorphism, chromosomal radiosensitivity and telomere length alterations closely resembling "Nijmegen breakage syndrome" phenotype. <i>European Journal of Medical Genetics</i> , 2007, 50, 176-187.	1.3	13

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91	The Role of microRNAs in the Biology of Rare Diseases. <i>International Journal of Molecular Sciences</i> , 2011, 12, 6733-6742.	4.1	13
92	The EuRRECa Project as a Model for Data Access and Governance Policies for Rare Disease Registries That Collect Clinical Outcomes. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 8743.	2.6	13
93	CPMSâ€“improving patient care in Europe via virtual case discussions. <i>Endocrine</i> , 2021, 71, 549-554.	2.3	13
94	Modeling delay to diagnosis for Amyotrophic lateral sclerosis: under reporting and incidence estimates. <i>BMC Neurology</i> , 2012, 12, 160.	1.8	12
95	Rare Diseases Research and Practice. <i>Endocrine Development</i> , 2014, 27, 234-256.	1.3	12
96	The Quality Evaluation of Rare Disease Registriesâ€”An Assessment of the Essential Features of a Disease Registry. <i>International Journal of Environmental Research and Public Health</i> , 2021, 18, 11968.	2.6	11
97	Primary Prevention of Congenital Anomalies: Special Focus on Environmental Chemicals and other Toxicants, Maternal Health and Health Services and Infectious Diseases. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 301-322.	1.6	10
98	Tackling rare diseases at European level: why do we need a harmonized framework?. <i>Folia Medica</i> , 2007, 49, 59-67.	0.5	10
99	Increased reactivity of laminin in the basement membranes of capillary walls in AIDS brain cortex. <i>Acta Neuropathologica</i> , 1991, 81, 552-556.	7.7	9
100	Eleven chromosomal integration sites of a human endogenous retrovirus (HERV 4-1) map close to known loci of thirteen hereditary malformation syndromes. <i>Teratology</i> , 1996, 54, 108-110.	1.6	9
101	The Role of Solidarity(-ies) in Rare Diseases Research. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 589-604.	1.6	9
102	Shaping the Future of Rare Diseases after a Global Health Emergency: Organisational Points to Consider. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 8694.	2.6	9
103	Shaping national plans and strategies for rare diseases in Europe: past, present, and future. <i>Journal of Community Genetics</i> , 2021, 12, 207-216.	1.2	9
104	Tackling the Problem of Rare Diseases in Public Health: The Italian Approach. <i>Public Health Genomics</i> , 2003, 6, 123-124.	1.0	8
105	Classification and codification of rare diseases. <i>Journal of Clinical Epidemiology</i> , 2012, 65, 1026-1027.	5.0	8
106	Recruitment procedures for descriptive socio-economic studies in rare diseases. The BURQOL-RD project. <i>Expert Opinion on Orphan Drugs</i> , 2015, 3, 759-765.	0.8	8
107	The Italian pilot external quality assessment program for cystic fibrosis sweat test. <i>Clinical Biochemistry</i> , 2016, 49, 601-605.	1.9	8
108	The Italian National Rare Diseases Registry: a model of comparison and integration with Hospital Discharge Data. <i>Journal of Public Health</i> , 2019, 41, 46-54.	1.8	8

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109	Reflections on the Importance of Cost of Illness Analysis in Rare Diseases: A Proposal. International Journal of Environmental Research and Public Health, 2021, 18, 1101.	2.6	8
110	Evaluation of an experimental periodontal ligament for dental implants. Biomaterials, 1991, 12, 474-478.	11.4	7
111	The Italian External Quality Control Programme for cystic fibrosis molecular diagnosis: 4 years of activity. Clinical Chemistry and Laboratory Medicine, 2007, 45, 254-60.	2.3	7
112	The Italian External Quality Assessment Scheme for Fragile X Syndrome: The Results of a 5-Year Survey. Genetic Testing and Molecular Biomarkers, 2008, 12, 279-288.	1.7	7
113	Availability of medicines for rare diseases in EU Countries. Pharmaceuticals Policy and Law, 2009, 11, 101-109.	0.1	7
114	Generation and characterization of a human chromosome 9 cosmid library. Somatic Cell and Molecular Genetics, 1992, 18, 269-284.	0.7	6
115	Increased c-met Expression During Ductal β^2 Cell Neogenesis in Experimental Autoimmune Diabetes. Growth Factors, 2001, 19, 259-267.	1.7	6
116	A new polymorphism in the flanking region of human VAMP2 and hPer1 genes. Molecular and Cellular Probes, 2002, 16, 391-392.	2.1	6
117	The Italian Scheme of External Quality Assessment for β^2 -Thalassemia: Genotyping and Reporting Results and Testing Strategies in a 5-Year Survey. Genetic Testing and Molecular Biomarkers, 2009, 13, 31-36.	0.7	6
118	Overview of existing initiatives to develop and improve access and data sharing in rare disease registries and biobanks worldwide. Expert Opinion on Orphan Drugs, 2016, 4, 729-739.	0.8	6
119	Social Economic Costs, Health-Related Quality of Life and Disability in Patients with Cri Du Chat Syndrome. International Journal of Environmental Research and Public Health, 2020, 17, 5951.	2.6	6
120	The Italian External Quality Assessment Scheme in Classical Cytogenetics: Four Years of Activity. Public Health Genomics, 2008, 11, 295-303.	1.0	5
121	Primary prevention as an essential factor ensuring sustainability of health systems: the example of congenital anomalies. Annali Dell'Istituto Superiore Di Sanita, 2019, 55, 258-264.	0.4	5
122	Expanded newborn bloodspot screening: developed country examples and what can be done in Turkey. Intractable and Rare Diseases Research, 2022, 11, 63-69.	0.9	5
123	Human endogenous retroviruses and environmental endocrine disruptors: A connection worth exploring?. , 1998, 58, 27-28.		4
124	The Italian National External Quality Assessment Program in Molecular Genetic Testing: Results of the VII Round (2010-2011). BioMed Research International, 2013, 2013, 1-8.	1.9	4
125	European Reference Networks and Guideline Development and Use: Challenges and Opportunities. Public Health Genomics, 2015, 18, 318-320.	1.0	4
126	The Italian National Centre for Rare Diseases: where research and public health translate into action. Blood Transfusion, 2014, 12 Suppl 3, s591-605.	0.4	4

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127	Europe and the century of biomedical discovery and implementation. <i>Lancet, The</i> , 2011, 377, 719-720.	13.7	3
128	Predictive medicine and biomarkers: the case of rare diseases. <i>Personalized Medicine</i> , 2012, 9, 143-146.	1.5	3
129	Molecular link(s) between hepatoblastoma pathogenesis and exposure to di-(2-ethylhexyl)phthalate: a hypothesis. <i>Folia Medica</i> , 2008, 50, 17-23.	0.5	3
130	The Italian External Quality Control Program for Familial Adenomatous Polyposis of the Colon: Five Years of Experience. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 175-181.	0.7	2
131	Health Systems Sustainability and Rare Diseases. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 629-640.	1.6	2
132	The Italian External Quality Assessment Program for Cystic Fibrosis Sweat Chloride Test: Does Active Participation Improve the Quality?. <i>International Journal of Environmental Research and Public Health</i> , 2020, 17, 3196.	2.6	2
133	Multifactorial Rare Diseases: Can Uncertainty Analysis Bring Added Value to the Search for Risk Factors and Etiopathogenesis?. <i>Medicina (Lithuania)</i> , 2021, 57, 119.	2.0	2
134	Sustainable public health systems for rare diseases. <i>Annali Dell'Istituto Superiore Di Sanita</i> , 2017, 53, 170-175.	0.4	2
135	Parent training education program: a pilot study, involving families of children with Prader-Willi syndrome. <i>Annali Dell'Istituto Superiore Di Sanita</i> , 2016, 52, 428-433.	0.4	2
136	Streptococcus Suis: A Potential Risk Factor for Salivary Gland Tumors?. <i>International Journal of Biological Markers</i> , 2011, 26, 278-280.	1.8	1
137	Modeling delay in diagnosis of NF: under reporting, incidence and prevalence estimates. <i>Journal of Applied Statistics</i> , 2012, 39, 711-721.	1.3	1
138	Editorial. <i>Public Health Genomics</i> , 2013, 16, 257-258.	1.0	1
139	Characterization and classification of Rare Disease Registries by using exploratory data analyses. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, P4.	2.7	1
140	Expanded Newborn Screening: A Chess Board Motif in Public Health. <i>Journal of Pediatric Biochemistry</i> , 2016, 06, 066-070.	0.2	1
141	The Genomic Medicine Alliance: A Global Effort to Facilitate the Introduction of Genomics into Healthcare in Developing Nations. , 2018, , 173-188.		1
142	Congenital anomalies: Can One Health reduce the community burden?. <i>Reproductive Toxicology</i> , 2021, 104, 166-167.	2.9	1
143	National Plans on Rare Diseases. <i>Advances in Predictive, Preventive and Personalised Medicine</i> , 2015, , 1-22.	0.6	1
144	Technologies to Support Frailty, Disability, and Rare Diseases: Towards a Monitoring Experience during the COVID-19 Pandemic Emergency. <i>Healthcare (Switzerland)</i> , 2022, 10, 235.	2.0	1

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145	Ultrastructure of the absorptive cell glycocalyx in hyperplastic colonic polyps after staining with Alcian Blue and high iron diamine. <i>The Histochemical Journal</i> , 1989, 21, 285-288.	0.6	0
146	PstI RFLP in the GABA _A receptor gene on human chromosome 6q. <i>Molecular and Cellular Probes</i> , 1997, 11, 309-310.	2.1	0
147	Quality of Life Assessment in Prader-Willi Syndrome. , 2011, , 3153-3162.		0
148	Haemophilia Centre Certification Systems: optional or optimal choice for healthcare systems?. <i>Blood Transfusion</i> , 2014, 12 Suppl 3, s492-4.	0.4	0
149	COVID-19 and the rare disease organization response during pandemic: the "Italian model". <i>Future Rare Diseases</i> , 2022, 2, .	0.4	0
150	Patient-physician alliance: from Hippocrates to Post-Genomic Era. Commentary. <i>Annali Dell'Istituto Superiore Di Sanita</i> , 2017, 53, 93-95.	0.4	0