Alessandro Mussa

List of Publications by Year
in descending order

131	3,720	117453	155451
papers	citations	34 h-index	55 g-index

133	133	133	3444
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genotypes and phenotypes heterogeneity in PIK3CA-related overgrowth spectrum and overlapping conditions: 150 novel patients and systematic review of 1007 patients with PIK3CA pathogenetic variants. Journal of Medical Genetics, 2023, 60, 163-173.	1.5	15
2	AKT/mTOR and MAPK Inhibition Improves Childhood RASopathic Cardiomyopathy. Thoracic and Cardiovascular Surgeon, 2022, 70, .	0.4	0
3	Maxillo-Facial Morphology in Beckwith-Wiedemann Syndrome: A Preliminary Study on (epi)Genotype-Phenotype Association in Caucasians. International Journal of Environmental Research and Public Health, 2022, 19, 2448.	1.2	7
4	MEK Inhibition in a Newborn with RAF1-Associated Noonan Syndrome Ameliorates Hypertrophic Cardiomyopathy but Is Insufficient to Revert Pulmonary Vascular Disease. Genes, 2022, 13, 6.	1.0	20
5	Epilepsy in a cohort of children with Noonan syndrome and related disorders. European Journal of Pediatrics, 2022, 181, 2919-2926.	1.3	4
6	Lateralized overgrowth with vascular malformation caused by a somatic PTPN11 pathogenic variant: Another piece added to the puzzle of mosaic <scp>RASopathies</scp>. Genes Chromosomes and Cancer, 2022, 61, 689-695.	1.5	4
7	Clinical and molecular characterization of patients affected by <scp>Beckwithâ€Wiedemann</scp> spectrum conceived through assisted reproduction techniques. Clinical Genetics, 2022, 102, 314-323.	1.0	7
8	Prenatal features in Beckwith-Wiedemann syndrome and indications for prenatal testing. Journal of Medical Genetics, 2021, 58, 842-849.	1.5	13
9	A new case of <scp>Smithâ€Kingsmore</scp> syndrome with somatic <scp>MTOR</scp> pathogenic variant expands the phenotypic spectrum to lateralized overgrowth. Clinical Genetics, 2021, 99, 719-723.	1.0	7
10	Atypical microdeletion 22q11.2 in a patient with tetralogy of Fallot. Journal of Genetics, 2021, 100, 1.	0.4	4
11	Wilms tumour occurring in a patient with osteopathia striata with cranial sclerosis: A still unsolved biological question. Pediatric Blood and Cancer, 2021, 68, e29132.	0.8	1
12	â€œThyroid nodular disease and PTEN mutation in a multicentre series of children with PTEN hamartoma tumor syndrome (PHTS)â€• Endocrine, 2021, 74, 632-637.	1.1	6
13	Evolution over Time of Leg Length Discrepancy in Patients with Syndromic and Isolated Lateralized Overgrowth. Journal of Pediatrics, 2021, 234, 123-127.	0.9	10
14	Correspondence on â€œClinical spectrum of MTOR-related hypomelanosis of Ito with neurodevelopmental abnormalities,â€•by Carmignac et al.. Genetics in Medicine, 2021, 23, 2223-2224.	1.1	1
15	Psychopathology and Adaptive Functioning in Children, Adolescents, and Young Adults with Noonan Syndrome. Journal of Developmental and Behavioral Pediatrics, 2021, Publish Ahead of Print, .	0.6	4
16	Kaposiform hemangioendothelioma further broadens the phenotype of <scp>PIK3CA</scp>â€related overgrowth spectrum. Clinical Genetics, 2021, 100, 624-627.	1.0	10
17	Treatment of Congenital Hypothyroidism: Comparison Between L-Thyroxine Oral Solution and Tablet Formulations up to 3 years of age. European Journal of Endocrinology, 2021, 186, 45-52.	1.9	4
18	Growth in Children With Noonan Syndrome and Effects of Growth Hormone Treatment on Adult Height. Frontiers in Endocrinology, 2021, 12, 761171.	1.5	6

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19	Lateralized and Segmental Overgrowth in Children. Cancers, 2021, 13, 6166.	1.7	10
20	Chronic subdural hematoma: A previously unreported lifeâ€threatening complication in adult with Sotos syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 3052-3055.	0.7	2
21	Syndromic Disorders Caused by Disturbed Human Imprinting. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 1-16.	0.4	24
22	Beckwith-Wiedemann Syndrome Negligible Effects on Tumor Risk Associated With In Vitro Fertilization. JAMA Pediatrics, 2019, 173, 996.	3.3	0
23	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.	1.9	1
24	Phenotype evolution and health issues of adults with Beckwithâ€Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1691-1702.	0.7	21
25	The effectiveness of Wilms tumor screening in Beckwithâ€“Wiedemann spectrum. Journal of Cancer Research and Clinical Oncology, 2019, 145, 3115-3123.	1.2	25
26	Cover Image, Volume 179A, Number 9, September 2019. , 2019, 179, i-i.		2
27	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	1.1	11
28	Longitudinal Monitoring of Alpha-Fetoprotein by Dried Blood Spot for Hepatoblastoma Screening in Beckwithâ€“Wiedemann Syndrome. Cancers, 2019, 11, 86.	1.7	7
29	Transcription alterations of KCNQ1 associated with imprinted methylation defects in the Beckwithâ€“Wiedemann locus. Genetics in Medicine, 2019, 21, 1808-1820.	1.1	38
30	Defining an optimal time window to screen for hepatoblastoma in children with Beckwithâ€Wiedemann syndrome. Pediatric Blood and Cancer, 2019, 66, e27492.	0.8	23
31	Revisiting Wilms tumour surveillance in Beckwithâ€“Wiedemann syndrome with IC2 methylation loss, reply. European Journal of Human Genetics, 2018, 26, 471-472.	1.4	13
32	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	4.3	388
33	Functional evaluation of natural killer cell cytotoxic activity in NFKB2-mutated patients. Immunology Letters, 2018, 194, 40-43.	1.1	12
34	Interaction between healthcare professionals and parents is a key determinant of parental distress during childhood hospitalisation for respiratory syncytial virus infection (European <scp>RSV</scp>) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 854-860.	0.7	17
35	Exposure to Gastric Acid Inhibitors Increases the Risk of Infection in Preterm Very Low Birth Weight Infants but Concomitant Administration of Lactoferrin Counteracts This Effect. Journal of Pediatrics, 2018, 193, 62-67.e1.	0.9	23
36	Assisted reproduction techniques and prenatal diagnosis of Beckwithâ€“Wiedemann spectrum presenting with omphalocele. Journal of Assisted Reproduction and Genetics, 2018, 35, 1925-1926.	1.2	1

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37	Serum alphaâ€fetoprotein screening for hepatoblastoma in Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 585-587.	0.7	10
38	Constitutional bone impairment in Noonan syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 692-698.	0.7	15
39	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	0.7	36
40	Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. Pediatrics, 2017, 140, .	1.0	87
41	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
42	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome: a paradigm for genomic medicine. Clinical Genetics, 2016, 89, 403-415.	1.0	57
43	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.	0.9	119
44	Fetal growth patterns in Beckwithâ€“Wiedemann syndrome. Clinical Genetics, 2016, 90, 21-27.	1.0	34
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.	1.8	54
46	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.	0.7	76
47	Genealogy of breastfeeding. European Journal of Pediatrics, 2016, 175, 105-112.	1.3	7
48	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	1.4	113
49	Predictivity of Clinical Findings and Doppler Ultrasound in Pediatric Acute Scrotum. Urology Journal, 2016, 13, 2779-83.	0.3	14
50	Screening Hepatoblastoma in Beckwith-Wiedemann Syndrome. Journal of Pediatric Hematology/Oncology, 2015, 37, 627.	0.3	31
51	Peculiarities of presentation and evolution over time of Hashimotoâ€™s thyroiditis in children and adolescents with Downâ€™s syndrome. Hormones, 2015, 14, 410-6.	0.9	33
52	Papillary thyroid cancer and autoimmune polyglandular syndrome. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 793-5.	0.4	8
53	The association with Turner syndrome significantly affects the course of Hashimotoâ€™s thyroiditis in children, irrespective of karyotype. Endocrine, 2015, 50, 777-782.	1.1	33
54	Predictors of Malignancy in Children with Thyroid Nodules. Journal of Pediatrics, 2015, 167, 886-892.e1.	0.9	73

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55	Levothyroxine requirement in congenital hypothyroidism: a 12-year longitudinal study. Endocrine, 2015, 50, 674-680.	1.1	12
56	Five-year prospective evaluation of thyroid function in girls with subclinical mild hypothyroidism of different etiology. European Journal of Endocrinology, 2015, 173, 801-808.	1.9	44
57	Underlying Hashimoto's Thyroiditis Negatively Affects the Evolution of Subclinical Hypothyroidism in Children Irrespective of Other Concomitant Risk Factors. Thyroid, 2015, 25, 183-187.	2.4	37
58	Thyroid Nodules and Carcinoma. , 2015, , 159-179.		1
59	The Complex Surgical Management of the First Case of Severe Combined Immunodeficiency and Multiple Intestinal Atresias Surviving after the Fourth Year of Life. Pediatric Gastroenterology, Hepatology and Nutrition, 2014, 17, 257.	0.4	9
60	Epidemiology, Presentation and Long-Term Evolution of Graves' Disease in Children, Adolescents and Young Adults with Turner Syndrome. Hormone Research in Paediatrics, 2014, 81, 245-250.	0.8	31
61	Iatrogenic acute pancreatitis due to hypercalcemia in a child with pseudohypoparathyroidism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 149-52.	0.4	3
62	Î±-Fetoprotein assay on dried blood spot for hepatoblastoma screening in children with overgrowth-cancer predisposition syndromes. Pediatric Research, 2014, 76, 544-548.	1.1	15
63	Fracture odds and body mass index in children. Journal of Pediatrics, 2014, 165, 1274.	0.9	4
64	Improved Sperm Count and Motility in Young Men Surgically Treated for Cryptorchidism in the First Year of Life. European Journal of Pediatric Surgery, 2014, 24, 376-380.	0.7	38
65	Successful medical treatment for ranula in children. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2014, 117, e289-e297.	0.2	9
66	Teriparatide (rhPTH) treatment in children with syndromic hypoparathyroidism. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 53-9.	0.4	16
67	Graves Disease in Children: Thyroid-Stimulating Hormone Receptor Antibodies as Remission Markers. Journal of Pediatrics, 2014, 164, 1189-1194.e1.	0.9	46
68	Subclinical hyperthyroidism when presenting as initial manifestation of juvenile Hashimotoâ€™s thyroiditis: first report on its natural history. Journal of Endocrinological Investigation, 2014, 37, 303-308.	1.8	22
69	Gastrostomy Intraperitoneal Bumper Migration in a Three-Year-Old Child: A Rare Complication following Gastrostomy Tube Replacement. Case Reports in Gastroenterology, 2014, 8, 381-386.	0.3	1
70	Phenotypic variability associated with the invariant SHOC2 c.4A>G (p.Ser2Gly) missense mutation. American Journal of Medical Genetics, Part A, 2014, 164, 3120-3125.	0.7	20
71	Prevalence of beckwithâ€“wiedemann syndrome in North West of Italy. American Journal of Medical Genetics, Part A, 2013, 161, 2481-2486.	0.7	93
72	Comment on â€œPrenatal diagnosis and prognosis in Noonan syndromeâ€• Prenatal Diagnosis, 2013, 33, 1318-1320.	1.1	3

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73	Serum Thyrotropin Concentration in Children with Isolated Thyroid Nodules. Journal of Pediatrics, 2013, 163, 1465-1470.	0.9	31
74	Later effects of metabolic control in phenylketonuria. Molecular Genetics and Metabolism, 2013, 109, 118.	0.5	0
75	Thyroid Involvement in Two Patients with Bannayan-Riley-Ruvalcaba Syndrome. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2013, 5, 261-265.	0.4	17
76	Central adrenal insufficiency in young adults with Praderâ€Willi Syndrome. Clinical Endocrinology, 2013, 79, 371-378.	1.2	29
77	Thyroid nodules in pediatrics: which ones can be left alone, which ones must be investigated, when and how. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 57-69.	0.4	25
78	Comparative Evaluation of Therapy with L-Thyroxine versus No Treatment in Children with Idiopathic and Mild Subclinical Hypothyroidism. Hormone Research in Paediatrics, 2012, 77, 376-381.	0.8	63
79	When to operate on ovarian cysts in children?. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 427-33.	0.4	12
80	Thyroid Function Patterns at Hashimotoâ€™s Thyroiditis Presentation in Childhood and Adolescence Are Mainly Conditioned by Patientsâ€™ Age. Hormone Research in Paediatrics, 2012, 78, 232-236.	0.8	64
81	Outcomes of Children with Hashitoxicosis. Hormone Research in Paediatrics, 2012, 77, 36-40.	0.8	56
82	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.	1.4	135
83	Polyuric-polydipsic syndrome in a pediatric case of non-glucocorticoid remediable familial hyperaldosteronism. Endocrine Journal, 2012, 59, 497-502.	0.7	24
84	Combined treatment with bicalutamide and anastrozole in a young boy with peripheral precocious puberty due to McCune-Albright Syndrome. Endocrine Journal, 2012, 59, 111-117.	0.7	20
85	Dopamine agonists in dihydropteridine reductase deficiency. Molecular Genetics and Metabolism, 2012, 105, 582-584.	0.5	15
86	Phalangeal quantitative ultrasound in 1,719 children and adolescents with bone disorders. Osteoporosis International, 2012, 23, 1987-1998.	1.3	9
87	The natural history of the normal/mild elevated TSH serum levels in children and adolescents with Hashimotoâ€™s thyroiditis and isolated hyperthyrotropinaemia: a 3â€year followâ€up. Clinical Endocrinology, 2012, 76, 394-398.	1.2	83
88	Assessment of central adrenal insufficiency in children and adolescents with Praderâ€“Willi syndrome. Clinical Endocrinology, 2012, 76, 843-850.	1.2	42
89	Nephrological findings and genotypeâ€“phenotype correlation in Beckwithâ€“Wiedemann syndrome. Pediatric Nephrology, 2012, 27, 397-406.	0.9	55
90	Lysosomal enzyme activities in phenylketonuria. Molecular Genetics and Metabolism, 2011, 102, 508-508.	0.5	2

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91	Phenotyping and treatment of phenylketonuria. Lancet, The, 2011, 377, 465.	6.3	5
92	Impact of Metabolic Control on Bone Quality in Phenylketonuria and Mild Hyperphenylalaninemia. Journal of Pediatric Gastroenterology and Nutrition, 2011, 52, 345-350.	0.9	15
93	Tetrahydrobiopterin and phenylketonuria. Journal of Pediatrics, 2011, 158, 864.	0.9	3
94	Determinants of thyrotropin rise in congenital hypothyroidism. Journal of Pediatrics, 2011, 159, 1050.	0.9	0
95	Neonatal hepatoblastoma in a newborn with severe phenotype of Beckwithâ€“Wiedemann syndrome. European Journal of Pediatrics, 2011, 170, 1407-1411.	1.3	38
96	Prenatal features of Noonan syndrome: prevalence and prognostic value. Prenatal Diagnosis, 2011, 31, 949-954.	1.1	43
97	Levothyroxine Treatment in Pediatric Benign Thyroid Nodules. Hormone Research in Paediatrics, 2011, 75, 246-251.	0.8	17
98	Surgical enucleation of testicular leydigioma in a young child: case report and literature review. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 839-42.	0.4	4
99	A heritable cause of cleft lip and palateâ€”Van der Woude syndrome caused by a novel IRF6 mutation. Review of the literature and of the differential diagnosis. European Journal of Pediatrics, 2010, 169, 223-228.	1.3	7
100	Remittent hyperammonemia in congenital portosystemic shunt. European Journal of Pediatrics, 2010, 169, 369-372.	1.3	18
101	Breastfeeding effects on newborn screening. Journal of Pediatrics, 2010, 156, 1033.	0.9	2
102	The overlap between Sotos and Beckwith-Wiedemann syndromes. Journal of Pediatrics, 2010, 156, 1035-1036.	0.9	2
103	Unresponsiveness to tetrahydrobiopterin of phenylalanine hydroxylase deficiency. Metabolism: Clinical and Experimental, 2010, 59, 645-652.	1.5	17
104	Bone Quantitative Ultrasound in Congenital and Acquired Childhood Multiple Pituitary Failure. Ultrasound in Medicine and Biology, 2010, 36, 726-732.	0.7	6
105	Management of sodium metabolism derangements in children treated for hypothalamic-hypophyseal tumors. Journal of Pediatric Biochemistry, 2010, 01, 289-296.	0.2	1
106	Frequency of Hashimotoâ€™s Thyroiditis Antecedents in the History of Children and Adolescents with Gravesâ€™ Disease. Hormone Research in Paediatrics, 2010, 73, 473-476.	0.8	45
107	Diagnostic Features of Thyroid Nodules in Pediatrics. JAMA Pediatrics, 2010, 164, 714.	3.6	79
108	Peculiarities of Graves' disease in children and adolescents with Down's syndrome. European Journal of Endocrinology, 2010, 162, 591-595.	1.9	55

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109	Gravesâ€™ disease prevalence in a young population with Turner syndrome. Journal of Endocrinological Investigation, 2010, 33, 69-70.	1.8	22
110	Prospective bone ultrasound patterns during childhood acute lymphoblastic leukemia treatment. Bone, 2010, 46, 1016-1020.	1.4	20
111	Fractures and skeletal complications should be the gold standard for validation of methods for bone appraisal in pediatrics. Bone, 2010, 47, 837-838.	1.4	2
112	Metabolic syndrome in children with Praderâ€“Willi syndrome: the effect of obesity. Nutrition, Metabolism and Cardiovascular Diseases, 2010, 21, 269-76.	1.1	46
113	Bone Impairment in Phenylketonuria Is Characterized by Circulating Osteoclast Precursors and Activated T Cell Increase. PLoS ONE, 2010, 5, e14167.	1.1	37
114	Sperm Count of Young Men Surgically Treated for Cryptorchidism in the First and Second Year of Life: Fertility is Better in Children Treated at a Younger Age. European Journal of Pediatric Surgery, 2009, 19, 388-391.	0.7	55
115	Dopamine agonists in 6-pyruvoyl tetrahydropterin synthase deficiency. Neurology, 2009, 73, 633-637.	1.5	32
116	Prospective evaluation of the natural course of idiopathic subclinical hypothyroidism in childhood and adolescence. European Journal of Endocrinology, 2009, 160, 417-421.	1.9	105
117	In response to van Spronsen et al (2009) Phenylalanine tolerance can already reliably be assessed at the age of 2 years in patients with PKU (J Inherit Metab Dis 32: 27â€“31). Journal of Inherited Metabolic Disease, 2009, 32, 584-586.	1.7	1
118	Increased spontaneous osteoclastogenesis from peripheral blood mononuclear cells in phenylketonuria. Journal of Inherited Metabolic Disease, 2008, 31, 339-342.	1.7	23
119	Phalangeal Quantitative Ultrasound in Children with Phenylketonuria: A Pilot Study. Ultrasound in Medicine and Biology, 2008, 34, 1049-1052.	0.7	20
120	Four new cases of PHACES syndrome: variable phenotypic expression and endocrine features. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 1729-1733.	0.7	4
121	Spontaneous osteoclastogenesis is a predictive factor for bone metastases from non-small cell lung cancer. Lung Cancer, 2008, 61, 109-116.	0.9	29
122	Thyroid Nodules and Cancer in Children and Adolescents Affected by Autoimmune Thyroiditis. JAMA Pediatrics, 2008, 162, 526.	3.6	116
123	Impact of Neonatal Protein Metabolism and Nutrition on Screening for Phenylketonuria. Journal of Pediatric Gastroenterology and Nutrition, 2008, 46, 561-569.	0.9	19
124	Congenital Hypothyroidism, Cerebellar Atrophy, and the Incomplete Phenotypic Expression of PHACES Syndrome. Endocrine Journal, 2008, 55, 231.	0.7	2
125	Bone Alterations in Children and Young Adults With Renal Transplant Assessed by Phalangeal Quantitative Ultrasound. American Journal of Kidney Diseases, 2007, 50, 441-449.	2.1	19
126	ILâ€7 Modulates Osteoclastogenesis in Patients Affected by Solid Tumors. Annals of the New York Academy of Sciences, 2007, 1117, 377-384.	1.8	20

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127	Goiter prevalence and urinary iodine status in urban and rural/mountain areas of Piedmont region. Journal of Endocrinological Investigation, 2006, 29, 67-73.	1.8	20
128	Image Diagnosis in McCune-Albright Syndrome. Journal of Pediatric Endocrinology and Metabolism, 2006, 19, 561-70.	0.4	13
129	Galectin-3 as a Presurgical Immunocytodiagnostic Marker of Minimally Invasive Follicular Thyroid Carcinoma. Journal of Clinical Endocrinology and Metabolism, 2001, 86, 5152-5158.	1.8	121
130	Intraoperative chemohyperthermia for advanced gastric cancer: a new procedure with closed abdomen and previously constructed anastomosis. Tumori, 2001, 87, S18-20.	0.6	3
131	Role of radioimmunolocalization in the staging of gastric carcinoma. , 1998, 15, 209-211.		3