

*PIK3CA*-related overgrowth spectrum (PROS):  
criteria, differential diagnosis, and evaluation

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Neurocutaneous Manifestations of Genetic Mosaicism. <i>Journal of Pediatric Genetics</i> , 2015, 04, 144-153.	0.3	8
2	Molecular and Functional Characterization of Three Different Postzygotic Mutations in PIK3CA-Related Overgrowth Spectrum (PROS) Patients: Effects on PI3K/AKT/mTOR Signaling and Sensitivity to PIK3 Inhibitors. <i>PLoS ONE</i> , 2015, 10, e0123092.	1.1	72
3	Molecular Genetics of Pediatric Orthopaedic Disorders. , 2015, , .		2
5	Update September 2015. <i>Lymphatic Research and Biology</i> , 2015, 13, 222-226.	0.5	0
6	Update March 2015. <i>Lymphatic Research and Biology</i> , 2015, 13, 66-73.	0.5	0
7	Clinical and Genetic Aspects of the Segmental Overgrowth Spectrum Due To Somatic Mutations in PIK3CA. <i>Journal of Pediatrics</i> , 2015, 167, 957-962.	0.9	29
9	Extending the spectrum of <i>AKT1</i> mosaicism: not just the Proteus syndrome. <i>British Journal of Dermatology</i> , 2016, 175, 612-614.	1.4	4
10	Nephroblastomatosis or Wilms tumor in a fourth patient with a somatic <i>PIK3CA</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2559-2569.	0.7	55
11	Phenotypic heterogeneity in <i>PIK3CA</i> -related overgrowth spectrum. <i>British Journal of Dermatology</i> , 2016, 175, 810-814.	1.4	10
12	Identification and Characterization of a Novel Constitutional <i>PIK3CA</i> Mutation in a Child Lacking the Typical Segmental Overgrowth of "PIK3CA-Related Overgrowth Spectrum" <i>Human Mutation</i> , 2016, 37, 242-245.	1.1	11
13	Somatic <i>PIK3CA</i> mutations as a driver of sporadic venous malformations. <i>Science Translational Medicine</i> , 2016, 8, 332ra42.	5.8	147
14	Phenotype with a side of genotype, please: Patients, parents and priorities in rare genetic disease. <i>Applied &amp; Translational Genomics</i> , 2016, 8, 42-44.	2.1	2
15	Vascular Diseases of the Spinal Cord: Infarction, Hemorrhage, and Venous Congestive Myelopathy. <i>Seminars in Ultrasound, CT and MRI</i> , 2016, 37, 466-481.	0.7	31
16	Spectrum of Fat-containing Soft-Tissue Masses at MR Imaging: The Common, the Uncommon, the Characteristic, and the Sometimes Confusing. <i>Radiographics</i> , 2016, 36, 753-766.	1.4	97
17	Tuberous Sclerosis Complex Associated with Vascular Anomalies or Overgrowth. <i>Pediatric Dermatology</i> , 2016, 33, 536-542.	0.5	18
18	Vascular Stains: Proposal for a Clinical Classification to Improve Diagnosis and Management. <i>Pediatric Dermatology</i> , 2016, 33, 570-584.	0.5	39
19	Klippel-Trenaunay syndrome belongs to the <i>PIK3CA</i> -related overgrowth spectrum (PROS). <i>Experimental Dermatology</i> , 2016, 25, 17-19.	1.4	143
20	Oral sildenafil as a treatment option for lymphatic malformations in <i>PIK3CA</i> -related tissue overgrowth syndromes. <i>Dermatologic Therapy</i> , 2016, 29, 466-469.	0.8	11

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21	Somatic overgrowth disorders of the PI3K/AKT/mTOR pathway & therapeutic strategies. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 402-421.	0.7	195
22	Somatic activating mutations in <i>Pik3ca</i> cause sporadic venous malformations in mice and humans. Science Translational Medicine, 2016, 8, 332ra43.	5.8	138
23	A Recurrent Mosaic Mutation in SMO , Encoding the Hedgehog Signal Transducer Smoothed, Is the Major Cause of Curry-Jones Syndrome. American Journal of Human Genetics, 2016, 98, 1256-1265.	2.6	70
25	De novo PIK3R2 variant causes polymicrogyria, corpus callosum hyperplasia and focal cortical dysplasia. European Journal of Human Genetics, 2016, 24, 1359-1362.	1.4	26
26	Overgrowth of the Hand and Upper Extremity and Associated Syndromes. Journal of Hand Surgery, 2016, 41, 473-482.	0.7	12
27	Molecular Genetics of the PI3K-AKT-mTOR Pathway in Genodermatoses: Diagnostic Implications and Treatment Opportunities. Journal of Investigative Dermatology, 2016, 136, 15-23.	0.3	35
28	Utility of clinical high-depth next generation sequencing for somatic variant detection in the <i>PIK3CA</i> -related overgrowth spectrum. Clinical Genetics, 2017, 91, 79-85.	1.0	40
29	Complex Truncal Masses in the Setting of CLOVES Syndrome: Aesthetic and Functional Implications. Aesthetic Plastic Surgery, 2017, 41, 591-599.	0.5	6
30	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	1.1	90
31	Localised intravascular coagulation complicating venous malformations in children: Associations and therapeutic options. Journal of Paediatrics and Child Health, 2017, 53, 737-741.	0.4	38
33	Fingertip Capillary Malformation and Associated Disorders: Report of 9 Cases. Pediatrics, 2017, 140, e20162967.	1.0	3
35	Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. European Journal of Endocrinology, 2017, 177, 175-186.	1.9	32
37	Somatic <i>PIK3CA</i> mutations in seven patients with <i>PIK3CA</i> -related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2017, 173, 978-984.	0.7	37
38	MANAGEMENT OF ENDOCRINE DISEASE: Diagnostic and therapeutic approach of tall stature. European Journal of Endocrinology, 2017, 176, R339-R353.	1.9	29
39	Advances in understanding and management of lymphoedema (cancer, primary). Current Opinion in Supportive and Palliative Care, 2017, 11, 355-360.	0.5	10
40	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	3.7	102
41	Exonic Mosaic Mutations Contribute Risk for Autism Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 369-390.	2.6	151
42	Prenatal Detection of PIK3CA-related Overgrowth Spectrum in Cultured Amniocytes Using Long-range PCR and Next-generation Sequencing. Pediatric and Developmental Pathology, 2017, 20, 54-57.	0.5	8

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43	Altered Adipose-Derived Stem Cell Characteristics in Macroductyly. Scientific Reports, 2017, 7, 11090.	1.6	6
44	Gait disturbance and lower limb pain in a patient with PIK3CA -related disorder. European Journal of Medical Genetics, 2017, 60, 655-657.	0.7	3
45	An approach to familial lymphoedema. Clinical Medicine, 2017, 17, 552-557.	0.8	20
46	Sonographic screening for Wilms tumor in children with CLOVES syndrome. Pediatric Blood and Cancer, 2017, 64, e26684.	0.8	52
48	Congenital Hemihyperplasia. , 2017, , 575-583.		0
49	Somatic PIK3CA mutations are present in multiple tissues of facial infiltrating lipomatosis. Pediatric Research, 2017, 82, 850-854.	1.1	28
50	<scp>CLOVES</scp> syndrome: review of a <scp>PIK3CA</scp>-related overgrowth spectrum (<scp>PROS</scp>). Clinical Genetics, 2017, 91, 14-21.	1.0	107
51	Nodular Proliferation in Parkes Weber Syndrome. Annals of Vascular Surgery, 2017, 38, 321.e1-321.e4.	0.4	1
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54	Human Mutations Associated With Brain Malformations Resulting in Hyperexcitability in Rodents. , 2017, , 827-844.		1
56	PI3K Signaling in Tissue Hyper-Proliferation: From Overgrowth Syndromes to Kidney Cysts. Cancers, 2017, 9, 30.	1.7	29
57	Venous Thromboembolism in Pediatric Vascular Anomalies. Frontiers in Pediatrics, 2017, 5, 158.	0.9	28
58	Identification of mutations in the PI3K-AKT-mTOR signalling pathway in patients with macrocephaly and developmental delay and/or autism. Molecular Autism, 2017, 8, 66.	2.6	85
59	The effect of rapamycin, NVP-BEZ235, aspirin, and metformin on PI3K/AKT/mTOR signaling pathway of <i>PIK3CA</i>-related overgrowth spectrum (PROS). Oncotarget, 2017, 8, 45470-45483.	0.8	17
60	An investigation of <i>PIK3CA</i> mutations in isolated macroductyly. Journal of Hand Surgery: European Volume, 2018, 43, 756-760.	0.5	22
61	Nonepithelial Tumors and Tumor-like Lesions of the Skin and Subcutis in Children. Pediatric and Developmental Pathology, 2018, 21, 150-207.	0.5	3
63	Structural malformations of the brain, eye, and pituitary gland in PHACE syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 48-55.	0.7	16
65	CLAPO syndrome: identification of somatic activating PIK3CA mutations and delineation of the natural history and phenotype. Genetics in Medicine, 2018, 20, 882-889.	1.1	52

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66	Syndromic Lymphedema and Complex Vascular Malformations with Lymphatic Involvement. , 2018, , 765-775.		0
67	PI3K/mTOR inhibition promotes the regression of experimental vascular malformations driven by PIK3CA-activating mutations. Cell Death and Disease, 2018, 9, 45.	2.7	59
68	Vascular Anomalies: From a Clinicohistologic to a Genetic Framework. Plastic and Reconstructive Surgery, 2018, 141, 709e-717e.	0.7	90
69	Analyzing the Genetic Spectrum of Vascular Anomalies with Overgrowth via Cancer Genomics. Journal of Investigative Dermatology, 2018, 138, 957-967.	0.3	45
70	New Frontiers in Our Understanding of Lymphatic Malformations of the Head and Neck. Otolaryngologic Clinics of North America, 2018, 51, 147-158.	0.5	37
71	Mosaic disorders and the Taxonomy of Human Disease. Genetics in Medicine, 2018, 20, 800-801.	1.1	6
72	In vitro efficacy of ARQ 092, an allosteric AKT inhibitor, on primary fibroblast cells derived from patients with PIK3CA-related overgrowth spectrum (PROS). Neurogenetics, 2018, 19, 77-91.	0.7	65
73	Scarring in Patients With PIK3CA-Related Overgrowth Syndromes. JAMA Dermatology, 2018, 154, 452.	2.0	13
74	Unilateral vestibular schwannoma and meningiomas in a patient with PIK3CA-related segmental overgrowth: Co-occurrence of mosaicism for 2 rare disorders. Clinical Genetics, 2018, 93, 187-190.	1.0	8
75	Treatment of Hand Macrodactyly With Resection and Toe Transfers. Journal of Hand Surgery, 2018, 43, 388.e1-388.e6.	0.7	10
76	Clinical and haemodynamic risk factors associated with discrepancies in lower limb length with capillary malformations: data from the national paediatric French cohort CONAPE. British Journal of Dermatology, 2018, 178, 520-526.	1.4	3
77	The importance of prenatal 3-dimensional sonography in a case of a segmental overgrowth syndrome with unclear chromosomal microarray results. Journal of Clinical Ultrasound, 2018, 46, 351-354.	0.4	0
78	Epidermal nevus syndromes: New insights into whorls and swirls. Pediatric Dermatology, 2018, 35, 21-29.	0.5	44
79	mTOR mutations in Smith-Kingsmore syndrome: Four additional patients and a review. Clinical Genetics, 2018, 93, 762-775.	1.0	36
80	Combined capillary-venous-lymphatic malformations without overgrowth in patients with Klippel-Tränaunay syndrome. Journal of Vascular Surgery: Venous and Lymphatic Disorders, 2018, 6, 230-236.	0.9	9
81	Causal somatic mutations in urine DNA from persons with the CLOVES subgroup of the PIK3CA-related overgrowth spectrum. Clinical Genetics, 2018, 93, 1075-1080.	1.0	20
82	Megalencephaly Capillary Malformation Syndrome. Journal of Pediatric Neurology, 2018, 16, 328-337.	0.0	6
83	Unilateral Type of Macrodystrophia Lipomatosa of the Thumb, Index Finger, and Thenar. Balkan Medical Journal, 2018, 36, 60-61.	0.3	1

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84	Phosphatidylinositol 3-Kinase, Growth Disorders, and Cancer. <i>New England Journal of Medicine</i> , 2018, 379, 2052-2062.	13.9	211
85	PHACE syndrome: Infantile hemangiomas associated with multiple congenital anomalies: Clues to the cause. , 2018, 178, 407-413.		22
86	Segmental epidermal nevus and mucosal neuromas associated with PIK3CA-related overgrowth spectrum disorder. <i>JAAD Case Reports</i> , 2018, 4, 1080-1082.	0.4	1
87	Clinical pitfalls in the diagnosis of segmental overgrowth syndromes: a child with the c.2740Gâ€%>â€%A mutation in PIK3CA gene. <i>Italian Journal of Pediatrics</i> , 2018, 44, 110.	1.0	5
88	Congenital infiltrating lipomatosis of the face with hyperplastic mandibular, maxillary and pterygoid bones: case report and a review of literature. <i>International Medical Case Reports Journal</i> , 2018, Volume 11, 233-238.	0.3	3
89	Characterization of the hepatosplenic and portal venous findings in patients with Proteus syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2677-2684.	0.7	4
90	Capillary Malformation. , 2018, , 149-159.		0
91	Upper limb muscle overgrowth with hypoplasia of the index finger: a new over-growth syndrome caused by the somatic PIK3CA mutation c.3140A&gt;G. <i>BMC Medical Genetics</i> , 2018, 19, 158.	2.1	11
92	An infant with a capillary malformation on the lower lip. <i>Pediatric Dermatology</i> , 2018, 35, 681-682.	0.5	0
93	Characterization of a severe case of <i>PIK3CA</i>â€related overgrowth at autopsy by droplet digital polymerase chain reaction and report of <i>PIK3CA</i> sequencing in 22 patients. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2301-2308.	0.7	20
94	Lessons for cancer drug treatment from tackling a non-cancerous overgrowth syndrome. <i>Nature</i> , 2018, 558, 523-525.	13.7	11
95	Polymicrogyria in association with hypoglycemia points to mutation in the mTOR pathway. <i>European Journal of Medical Genetics</i> , 2018, 61, 738-740.	0.7	12
96	Vascular Anomalies. <i>Journal of Hand Surgery</i> , 2018, 43, 1113-1121.	0.7	11
97	Targeted therapy in patients with PIK3CA-related overgrowth syndrome. <i>Nature</i> , 2018, 558, 540-546.	13.7	374
98	Pulmonary thromboembolic events in patients with congenital lipomatous overgrowth, vascular malformations, epidermal nevi, and spinal/skeletal abnormalities and Klippel-TrÃ©naunay syndrome. <i>Journal of Vascular Surgery: Venous and Lymphatic Disorders</i> , 2018, 6, 511-516.	0.9	39
99	A large shagreen patch with overlying verrucous epidermal naevus: a curious case of colocalization. <i>Clinical and Experimental Dermatology</i> , 2019, 44, 218-220.	0.6	1
100	Insights into the pathogenesis of macrodactyly. <i>Journal of Hand Surgery: European Volume</i> , 2019, 44, 25-31.	0.5	6
101	Molecular heterogeneity of the cerebriform connective tissue nevus in mosaic overgrowth syndromes. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a004036.	0.5	3

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102	A girl with CLOVES syndrome with a recurrent PIK3CA somatic mutation and pancreatic steatosis. <i>Human Genome Variation</i> , 2019, 6, 31.	0.4	4
103	Germline pathogenic variant in <i>PIK3CA</i> leading to symmetrical overgrowth with marked macrocephaly and mild global developmental delay. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e845.	0.6	11
104	Alpelisib Treatment for Genital Vascular Malformation in a Patient with Congenital Lipomatous Overgrowth, Vascular Malformations, Epidermal Nevi, and Spinal/Skeletal Anomalies and/or Scoliosis (CLOVES) Syndrome. <i>Journal of Pediatric and Adolescent Gynecology</i> , 2019, 32, 648-650.	0.3	43
105	A dyadic genotype-phenotype approach to diagnostic criteria for Proteus syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 565-570.	0.7	24
106	Thrombosis risk factors in <i>PIK3CA</i> -related overgrowth spectrum and Proteus syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 571-581.	0.7	28
107	Diagnostic Utility of Next-Generation Sequencing for Disorders of Somatic Mosaicism: A Five-Year Cumulative Cohort. <i>American Journal of Human Genetics</i> , 2019, 105, 734-746.	2.6	23
109	Generalized Lymphatic Anomaly and Gorham-Stout Disease: Overview and Recent Insights. <i>Advances in Wound Care</i> , 2019, 8, 230-245.	2.6	76
110	Efficacy of systemic sirolimus in the treatment of generalized lymphatic anomaly and Gorham-Stout disease. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27614.	0.8	81
111	Comprehensive molecular and clinicopathological analysis of vascular malformations: A study of 319 cases. <i>Genes Chromosomes and Cancer</i> , 2019, 58, 541-550.	1.5	50
112	Functional characterisation of a novel class of in-frame insertion variants of KRAS and HRAS. <i>Scientific Reports</i> , 2019, 9, 8239.	1.6	12
114	Old treatments for new genetic conditions: Sirolimus therapy in a child affected by mosaic overgrowth with fibroadipose hyperplasia. <i>Clinical Genetics</i> , 2019, 96, 102-103.	1.0	6
116	Signaling pathways and inhibitors of cells from patients with kaposiform lymphangiomatosis. <i>Pediatric Blood and Cancer</i> , 2019, 66, e27790.	0.8	18
117	Congenital Limb Overgrowth Syndromes Associated with Vascular Anomalies. <i>Radiographics</i> , 2019, 39, 491-515.	1.4	43
118	Clinical application of molecular genetics in lymphatic malformations. <i>Laryngoscope Investigative Otolaryngology</i> , 2019, 4, 170-173.	0.6	11
119	Early activating somatic <i>PIK3CA</i> mutations promote ectopic muscle development and upper limb overgrowth. <i>Clinical Genetics</i> , 2019, 96, 118-125.	1.0	14
120	Acute leukemia in a patient with 15q overgrowth syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1025-1029.	0.7	0
121	Laser ablation of embryonic veins in children. <i>Pediatrics International</i> , 2019, 61, 358-363.	0.2	9
122	Adipocytic tumors in Children: A contemporary review. <i>Seminars in Diagnostic Pathology</i> , 2019, 36, 95-104.	1.0	26

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123	Hormonal receptors in cutaneous vascular malformations: 51 cases. <i>Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin</i> , 2019, 474, 755-761.	1.4	7
124	Molecular diagnosis of somatic overgrowth conditions: A single-center experience. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e536.	0.6	28
127	Practical Genetic and Biologic Therapeutic Considerations in Vascular Anomalies. <i>Techniques in Vascular and Interventional Radiology</i> , 2019, 22, 100629.	0.4	10
128	Macrodactyly: decision-making and surgery timing. <i>Journal of Hand Surgery: European Volume</i> , 2019, 44, 32-42.	0.5	18
129	Somatic activating mutations in <i>PIK3CA</i> cause generalized lymphatic anomaly. <i>Journal of Experimental Medicine</i> , 2019, 216, 407-418.	4.2	96
130	CHAPLE syndrome uncovers the primary role of complement in a familial form of Waldmann's disease. <i>Immunological Reviews</i> , 2019, 287, 20-32.	2.8	18
131	Reply: "Developmental venous anomaly depicted incidentally in fetal MRI and confirmed in post-natal MRI". <i>Neuroradiology</i> , 2019, 61, 11-12.	1.1	3
132	A postzygotic KRAS mutation in a patient with Schimmelpenning syndrome presenting with lipomatosis, renovascular hypertension, and diabetes mellitus. <i>Journal of Human Genetics</i> , 2019, 64, 177-181.	1.1	14
133	Constitutional mosaicism in <i>RASA1</i> -related capillary malformation-arteriovenous malformation. <i>Clinical Genetics</i> , 2019, 95, 516-519.	1.0	10
134	A Child With Lymphangioma Due to Somatic Mutation in PIK3CA Successfully Treated With Everolimus. <i>Pediatric Neurology</i> , 2019, 91, 65-67.	1.0	2
135	Vascular anomalies of the upper limb. <i>Journal of Hand Surgery: European Volume</i> , 2019, 44, 233-241.	0.5	4
136	Abnormal Body Size and Proportion. , 2019, , 81-143.		0
137	Safety and efficacy of low-dose sirolimus in the PIK3CA-related overgrowth spectrum. <i>Genetics in Medicine</i> , 2019, 21, 1189-1198.	1.1	115
138	Mosaic abnormalities of the skin: review and guidelines from the European Reference Network for rare skin diseases. <i>British Journal of Dermatology</i> , 2020, 182, 552-563.	1.4	45
139	Novel features of PIK3CA-Related Overgrowth Spectrum: Lesson from an aborted fetus presenting a de novo constitutional PIK3CA mutation. <i>European Journal of Medical Genetics</i> , 2020, 63, 103775.	0.7	7
140	PIK3CA mutations in lipomatosis of nerve with or without nerve territory overgrowth. <i>Modern Pathology</i> , 2020, 33, 420-430.	2.9	33
141	Ultrasound-Mediated Gene Therapy of Hepatocellular Carcinoma Using Pre-microRNA Plasmid-Loaded Nanodroplets. <i>Ultrasound in Medicine and Biology</i> , 2020, 46, 90-107.	0.7	13
142	Genetic Disorders of the Lymphatic System. , 2020, , 231-249.		2



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143	Soft tissue angiomatosis: another PIK3CA related disorder. <i>Histopathology</i> , 2020, 76, 540-549.	1.6	12
144	A swollen cheek. <i>Journal of Stomatology, Oral and Maxillofacial Surgery</i> , 2020, 121, 461-462.	0.5	0
145	Diffuse capillary malformation with overgrowth contains somatic PIK3CA variants. <i>Clinical Genetics</i> , 2020, 97, 736-740.	1.0	22
146	Disorders of the Venous System. , 2020, , 251-260.		0
147	Reverse Phenotyping in Patients with Skin Capillary Malformations and Mosaic GNAQ or GNA11 Mutations Defines a Clinical Spectrum with Genotype-Phenotype Correlation. <i>Journal of Investigative Dermatology</i> , 2020, 140, 1106-1110.e2.	0.3	30
148	Prenatal imaging diagnosis of PIK3CA related overgrowth spectrum disorders in first trimester with emphasis on extremities. <i>Ultrasound in Obstetrics and Gynecology</i> , 2020, 56, 780-781.	0.9	4
149	Congenital enlargement of toes. <i>Pediatric Dermatology</i> , 2020, 37, 945-946.	0.5	0
150	Familial hypercholesterolemia: A single-nucleotide variant (SNV) in mosaic at the low density lipoprotein receptor (LDLR). <i>Atherosclerosis</i> , 2020, 311, 37-43.	0.4	5
151	Overgrowth syndromes and new therapies. <i>Seminars in Pediatric Surgery</i> , 2020, 29, 150974.	0.5	4
152	Orofacial overgrowth with peripheral nerve enlargement and perineuriomatous pseudo-onion bulb proliferations is part of the PIK3CA-related overgrowth spectrum. <i>Human Genetics and Genomics Advances</i> , 2020, 1, 100009.	1.0	6
153	A six-attribute classification of genetic mosaicism. <i>Genetics in Medicine</i> , 2020, 22, 1743-1757.	1.1	34
154	PIK3CA vascular overgrowth syndromes: an update. <i>Current Opinion in Pediatrics</i> , 2020, 32, 539-546.	1.0	32
155	Activating PIK3CA mutation promotes adipogenesis of adipose-derived stem cells in macrodactyly via up-regulation of E2F1. <i>Cell Death and Disease</i> , 2020, 11, 600.	2.7	7
156	Definitions and classification of malformations of cortical development: practical guidelines. <i>Brain</i> , 2020, 143, 2874-2894.	3.7	145
157	Detailed analysis of phenotypes and genotypes in megalencephaly-capillary malformation-polymicrogyria syndrome caused by somatic mosaicism of PIK3CA mutations. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 205.	1.2	14
158	Phenotypic and genetic spectrum of isolated macrodactyly: somatic mosaicism of PIK3CA and AKT1 oncogenic variants. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 288.	1.2	15
159	Newcomers in Vascular Anomalies. <i>Surgical Pathology Clinics</i> , 2020, 13, 719-728.	0.7	1
160	Ubiquitous expression of Akt1 p.(E17K) results in vascular defects and embryonic lethality in mice. <i>Human Molecular Genetics</i> , 2020, 29, 3350-3360.	1.4	1

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161	Genomically Aided Diagnosis of Severe Developmental Disorders. Annual Review of Genomics and Human Genetics, 2020, 21, 327-349.	2.5	3
162	Disease-associated mosaic variation in clinical exome sequencing: a two-year pediatric tertiary care experience. Journal of Physical Education and Sports Management, 2020, 6, a005231.	0.5	15
163	Update and audit of the St George's™s classification algorithm of primary lymphatic anomalies: a clinical and molecular approach to diagnosis. Journal of Medical Genetics, 2020, 57, 653-659.	1.5	59
164	Brain morphological analysis in PTEN hamartoma tumor syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1117-1129.	0.7	12
165	Expanding the phenotypic spectrum of lipomatosis of the sciatic nerve: Early-onset colonic diverticular disease. Neurogastroenterology and Motility, 2020, 32, e13917.	1.6	2
166	PIK3CA variants selectively initiate brain hyperactivity during gliomagenesis. Nature, 2020, 578, 166-171.	13.7	131
167	Constitutively active PIK3CA mutations are expressed by lymphatic and vascular endothelial cells in capillary lymphatic venous malformation. Angiogenesis, 2020, 23, 425-442.	3.7	34
168	A 34-Year-Old Man With a Chylothorax and Bony Pain. Chest, 2020, 157, e131-e136.	0.4	1
169	Theranostic Advances in Vascular Malformations. Journal of Investigative Dermatology, 2020, 140, 756-763.	0.3	41
170	CD10 and CD34 as markers in vascular malformations with PIK3CA and TEK mutations. Human Pathology, 2020, 99, 98-106.	1.1	2
171	A pilot study of next generation sequencing—liquid biopsy on cell-free DNA as a novel non-invasive diagnostic tool for Klippel-Trenaunay syndrome. Vascular, 2021, 29, 85-91.	0.4	14
172	Genetic control of tumor development in malformation syndromes. American Journal of Medical Genetics, Part A, 2021, 185, 324-335.	0.7	2
173	Lipoblastoma phenotype contains a somatic PIK3CA mutation. Pediatric Dermatology, 2021, 38, 299-300.	0.5	3
174	Asymmetric Overgrowth and a Facial Port Wine Stain. Journal of Pediatrics, 2021, 229, 300-301.	0.9	0
175	Vascular anomalies: Classification and management. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 176, 345-360.	1.0	3
176	Custom Pediatric Oncology Next-Generation Sequencing Panel Identifies Somatic Mosaicism in Archival Tissue and Enhances Targeted Clinical Care. Pediatric Neurology, 2021, 114, 55-59.	1.0	1
177	Mutant Allele Imbalance in Cancer. Annual Review of Cancer Biology, 2021, 5, 221-234.	2.3	2
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357	Successful Treatment of Hypoglycemia With Alpelisib in Pediatric Patients With <i>PIK3CA</i> -Related Overgrowth Spectrum. , 2023, 1, .		1
364	Brain Malformations. , 2024, , 61-166.		0
366	Traitement des malformations vasculaires pŕiphŕiques et formes complexes. , 2023, , 317-325.		0
368	Epidemiology of the disorders of the <i>Pik3ca</i> -related overgrowth spectrum (Pros). <i>European Journal of Human Genetics</i> , 0, , .	1.4	3
375	Case Report: Sequential postzygotic <i>HRAS</i> mutation and gains of the paternal chromosome 11 carrying the mutated allele in a patient with epidermal nevus and rhabdomyosarcoma: evidence of a multiple-hit mechanism involving <i>HRAS</i> in oncogenic transformation. <i>Frontiers in Genetics</i> , 0, 14, .	1.1	0
383	Macrodactilies. , 2023, , 269-281.		0
392	The expanding diagnostic toolbox for rare genetic diseases. <i>Nature Reviews Genetics</i> , 0, , .	7.7	0
394	Progressive vascular tumor in infant: A case report and literature review of <i>PIK3CA</i> vascular malformation. <i>Child's Nervous System</i> , 2024, 40, 1005-1010.	0.6	0