

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. Journal of Pediatric Genetics, 2015, 04, 144-153.	0.7	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. PLoS ONE, 2015, 10, e0123092.	2.5	72
3	Molecular Genetics of Pediatric Orthopaedic Disorders. , 2015, , .		2
5	Update September 2015. Lymphatic Research and Biology, 2015, 13, 222-226.	1.1	0
6	Update March 2015. Lymphatic Research and Biology, 2015, 13, 66-73.	1.1	0
7	Clinical and Genetic Aspects of the Segmental Overgrowth Spectrum DueÂto Somatic Mutations in PIK3CA. Journal of Pediatrics, 2015, 167, 957-962.	1.8	29
9	Extending the spectrum of<i>AKT1</i> mosaicism: not just the Proteus syndrome. British Journal of Dermatology, 2016, 175, 612-614.	1.5	4
10	Nephroblastomatosis or Wilms tumor in a fourth patient with a somatic <i>PIK3CA</i> mutation. American Journal of Medical Genetics, Part A, 2016, 170, 2559-2569.	1.2	55
11	Phenotypic heterogeneity inPIK3CA-related overgrowth spectrum. British Journal of Dermatology, 2016, 175, 810-814.	1.5	10
12	Identification and Characterization of a Novel ConstitutionalPIK3CAMutation in a Child Lacking the Typical Segmental Overgrowth of â€œPIK3CA-Related Overgrowth Spectrumâ€ Human Mutation, 2016, 37, 242-245.	2.5	11
13	Somatic <i>PIK3CA</i> mutations as a driver of sporadic venous malformations. Science Translational Medicine, 2016, 8, 332ra42.	12.4	147
14	Phenotype with a side of genotype, please: Patients, parents and priorities in rare genetic disease. Applied & Translational Genomics, 2016, 8, 42-44.	2.1	2
15	Vascular Diseases of the Spinal Cord: Infarction, Hemorrhage, and Venous Congestive Myelopathy. Seminars in Ultrasound, CT and MRI, 2016, 37, 466-481.	1.5	31
16	Spectrum of Fat-containing Soft-Tissue Masses at MR Imaging: The Common, the Uncommon, the Characteristic, and the Sometimes Confusing. Radiographics, 2016, 36, 753-766.	3.3	97
17	Tuberous Sclerosis Complex Associated with Vascular Anomalies or Overgrowth. Pediatric Dermatology, 2016, 33, 536-542.	0.9	18
18	Vascular Stains: Proposal for a Clinical Classification to Improve Diagnosis and Management. Pediatric Dermatology, 2016, 33, 570-584.	0.9	39
19	Klippelâ€Trenaunay syndrome belongs to the <i>PIK3CA</i>â€related overgrowth spectrum (PROS). Experimental Dermatology, 2016, 25, 17-19.	2.9	143
20	Oral sildenafil as a treatment option for lymphatic malformations in PIK3CA-related tissue overgrowth syndromes. Dermatologic Therapy, 2016, 29, 466-469.	1.7	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.	1.6	195
22	Somatic activating mutations in <i>Pik3ca</i> cause sporadic venous malformations in mice and humans. Science Translational Medicine, 2016, 8, 332ra43.	12.4	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.	6.2	70
25	De novo PIK3R2 variant causes polymicrogyria, corpus callosum hyperplasia and focal cortical dysplasia. European Journal of Human Genetics, 2016, 24, 1359-1362.	2.8	26
26	Overgrowth of the Hand and Upper Extremity and Associated Syndromes. Journal of Hand Surgery, 2016, 41, 473-482.	1.6	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.	0.7	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.	2.0	40
29	Complex Truncal Masses in the Setting of CLOVES Syndrome: Aesthetic and Functional Implications. Aesthetic Plastic Surgery, 2017, 41, 591-599.	0.9	6
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31	Localised intravascular coagulation complicating venous malformations in children: Associations and therapeutic options. Journal of Paediatrics and Child Health, 2017, 53, 737-741.	0.8	38
33	Fingertip Capillary Malformation and Associated Disorders: Report of 9 Cases. Pediatrics, 2017, 140, e20162967.	2.1	3
35	Hypoinsulinaemic, hypoketotic hypoglycaemia due to mosaic genetic activation of PI3-kinase. European Journal of Endocrinology, 2017, 177, 175-186.	3.7	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.	1.2	37
38	MANAGEMENT OF ENDOCRINE DISEASE: Diagnostic and therapeutic approach of tall stature. European Journal of Endocrinology, 2017, 176, R339-R353.	3.7	29
39	Advances in understanding and management of lymphoedema (cancer, primary). Current Opinion in Supportive and Palliative Care, 2017, 11, 355-360.	1.3	10
40	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
41	Exonic Mosaic Mutations Contribute Risk for Autism Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 369-390.	6.2	151
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44	Gait disturbance and lower limb pain in a patient with PIK3CA -related disorder. European Journal of Medical Genetics, 2017, 60, 655-657.	1.3	3
45	An approach to familial lymphoedema. Clinical Medicine, 2017, 17, 552-557.	1.9	20
46	Sonographic screening for Wilms tumor in children with CLOVES syndrome. Pediatric Blood and Cancer, 2017, 64, e26684.	1.5	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.	2.3	28
50	<scp>CLOVES</scp> syndrome: review of a <scp>PIK3CA</scp>-related overgrowth spectrum (<scp>PROS</scp>). Clinical Genetics, 2017, 91, 14-21.	2.0	107
51	Nodular Proliferation in Parkes Weber Syndrome. Annals of Vascular Surgery, 2017, 38, 321.e1-321.e4.	0.9	1
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56	PI3K Signaling in Tissue Hyper-Proliferation: From Overgrowth Syndromes to Kidney Cysts. Cancers, 2017, 9, 30.	3.7	29
57	Venous Thromboembolism in Pediatric Vascular Anomalies. Frontiers in Pediatrics, 2017, 5, 158.	1.9	28
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60	An investigation of <i>PIK3CA</i> mutations in isolated macroductyly. Journal of Hand Surgery: European Volume, 2018, 43, 756-760.	1.0	22
61	Nonepithelial Tumors and Tumor-like Lesions of the Skin and Subcutis in Children. Pediatric and Developmental Pathology, 2018, 21, 150-207.	1.0	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.	1.2	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.	2.4	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.	6.3	59
68	Vascular Anomalies: From a Clinicohistologic to a Genetic Framework. Plastic and Reconstructive Surgery, 2018, 141, 709e-717e.	1.4	90
69	Analyzing the Genetic Spectrum of Vascular Anomalies with Overgrowth via Cancer Genomics. Journal of Investigative Dermatology, 2018, 138, 957-967.	0.7	45
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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.	1.5	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.	0.8	0
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79	mTOR mutations in Smith-Kingsmore syndrome: Four additional patients and a review. Clinical Genetics, 2018, 93, 762-775.	2.0	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.	1.6	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.	2.0	20
82	Megalencephaly Capillary Malformation Syndrome. Journal of Pediatric Neurology, 2018, 16, 328-337.	0.2	6
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85	PHACE syndrome: Infantile hemangiomas associated with multiple congenital anomalies: Clues to the cause. , 2018, 178, 407-413.		22
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88	Congenital infiltrating lipomatosis of the face with hyperplastic mandibular, maxillary and pterygoid bones: case report and a review of literature. International Medical Case Reports Journal, 2018, Volume 11, 233-238.	0.8	3
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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>G. BMC Medical Genetics, 2018, 19, 158.	2.1	11
92	An infant with a capillary malformation on the lower lip. Pediatric Dermatology, 2018, 35, 681-682.	0.9	0
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99	A large shagreen patch with overlying verrucous epidermal naevus: a curious case of colocalization. Clinical and Experimental Dermatology, 2019, 44, 218-220.	1.3	1
100	Insights into the pathogenesis of macrodactyly. Journal of Hand Surgery: European Volume, 2019, 44, 25-31.	1.0	6
101	Molecular heterogeneity of the cerebriform connective tissue nevus in mosaic overgrowth syndromes. Journal of Physical Education and Sports Management, 2019, 5, a004036.	1.2	3

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102	A girl with CLOVES syndrome with a recurrent PIK3CA somatic mutation and pancreatic steatosis. Human Genome Variation, 2019, 6, 31.	0.7	4
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110	Efficacy of systemic sirolimus in the treatment of generalized lymphatic anomaly and Gorham-Stout disease. Pediatric Blood and Cancer, 2019, 66, e27614.	1.5	81
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118	Clinical application of molecular genetics in lymphatic malformations. Laryngoscope Investigative Otolaryngology, 2019, 4, 170-173.	1.5	11
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131	Reply: "Developmental venous anomaly depicted incidentally in fetal MRI and confirmed in post-natal MRI". Neuroradiology, 2019, 61, 11-12.	2.2	3
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133	Constitutional mosaicism in <i>RASA1</i> -related capillary malformation-arteriovenous malformation. Clinical Genetics, 2019, 95, 516-519.	2.0	10
134	A Child With Lymphangioma Due to Somatic Mutation in PIK3CA Successfully Treated With Everolimus. Pediatric Neurology, 2019, 91, 65-67.	2.1	2
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138	Mosaic abnormalities of the skin: review and guidelines from the European Reference Network for rare skin diseases. British Journal of Dermatology, 2020, 182, 552-563.	1.5	45
139	Novel features of PIK3CA-Related Overgrowth Spectrum: Lesson from an aborted fetus presenting a de novo constitutional PIK3CA mutation. European Journal of Medical Genetics, 2020, 63, 103775.	1.3	7
140	PIK3CA mutations in lipomatosis of nerve with or without nerve territory overgrowth. Modern Pathology, 2020, 33, 420-430.	5.5	33
141	Ultrasound-Mediated Gene Therapy of Hepatocellular Carcinoma Using Pre-microRNA Plasmid-Loaded Nanodroplets. Ultrasound in Medicine and Biology, 2020, 46, 90-107.	1.5	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.	2.9	12
144	A swollen cheek. <i>Journal of Stomatology, Oral and Maxillofacial Surgery</i> , 2020, 121, 461-462.	1.3	0
145	Diffuse capillary malformation with overgrowth contains somatic <i>PIK3CA</i> variants. <i>Clinical Genetics</i> , 2020, 97, 736-740.	2.0	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.	0.7	30
148	Prenatal imaging diagnosis of <i>PIK3CA</i> -related overgrowth spectrum disorders in first trimester with emphasis on extremities. <i>Ultrasound in Obstetrics and Gynecology</i> , 2020, 56, 780-781.	1.7	4
149	Congenital enlargement of toes. <i>Pediatric Dermatology</i> , 2020, 37, 945-946.	0.9	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.	0.8	5
151	Overgrowth syndromes and new therapies. <i>Seminars in Pediatric Surgery</i> , 2020, 29, 150974.	1.1	4
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153	A six-attribute classification of genetic mosaicism. <i>Genetics in Medicine</i> , 2020, 22, 1743-1757.	2.4	34
154	<i>PIK3CA</i> vascular overgrowth syndromes: an update. <i>Current Opinion in Pediatrics</i> , 2020, 32, 539-546.	2.0	32
155	Activating <i>PIK3CA</i> mutation promotes adipogenesis of adipose-derived stem cells in macrodactyly via up-regulation of E2F1. <i>Cell Death and Disease</i> , 2020, 11, 600.	6.3	7
156	Definitions and classification of malformations of cortical development: practical guidelines. <i>Brain</i> , 2020, 143, 2874-2894.	7.6	145
157	Detailed analysis of phenotypes and genotypes in megalencephaly-capillary malformation-polymicrogyria syndrome caused by somatic mosaicism of <i>PIK3CA</i> mutations. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 205.	2.7	14
158	Phenotypic and genetic spectrum of isolated macrodactyly: somatic mosaicism of <i>PIK3CA</i> and <i>AKT1</i> oncogenic variants. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 288.	2.7	15
159	Newcomers in Vascular Anomalies. <i>Surgical Pathology Clinics</i> , 2020, 13, 719-728.	1.7	1
160	Ubiquitous expression of <i>Akt1</i> p.(E17K) results in vascular defects and embryonic lethality in mice. <i>Human Molecular Genetics</i> , 2020, 29, 3350-3360.	2.9	1

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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.	1.2	15
163	Update and audit of the St George's™ classification algorithm of primary lymphatic anomalies: a clinical and molecular approach to diagnosis. Journal of Medical Genetics, 2020, 57, 653-659.	3.2	59
164	Brain morphological analysis in PTEN hamartoma tumor syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1117-1129.	1.2	12
165	Expanding the phenotypic spectrum of lipomatosis of the sciatic nerve: Early-onset colonic diverticular disease. Neurogastroenterology and Motility, 2020, 32, e13917.	3.0	2
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167	Constitutively active PIK3CA mutations are expressed by lymphatic and vascular endothelial cells in capillary lymphatic venous malformation. Angiogenesis, 2020, 23, 425-442.	7.2	34
168	A 34-Year-Old Man With a Chylothorax and Bony Pain. Chest, 2020, 157, e131-e136.	0.8	1
169	Theranostic Advances in Vascular Malformations. Journal of Investigative Dermatology, 2020, 140, 756-763.	0.7	41
170	CD10 and CD34 as markers in vascular malformations with PIK3CA and TEK mutations. Human Pathology, 2020, 99, 98-106.	2.0	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.	0.9	14
172	Genetic control of tumor development in malformation syndromes. American Journal of Medical Genetics, Part A, 2021, 185, 324-335.	1.2	2
173	Lipoblastoma phenotype contains a somatic PIK3CA mutation. Pediatric Dermatology, 2021, 38, 299-300.	0.9	3
174	Asymmetric Overgrowth and a Facial Port Wine Stain. Journal of Pediatrics, 2021, 229, 300-301.	1.8	0
175	Vascular anomalies: Classification and management. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2021, 176, 345-360.	1.8	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.	2.1	1
177	Mutant Allele Imbalance in Cancer. Annual Review of Cancer Biology, 2021, 5, 221-234.	4.5	2
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180	Pathogenic postzygotic mosaicism in the tyrosine receptor kinase pathway: potential unidentified human disease hidden away in a few cells. FEBS Journal, 2021, 288, 3108-3119.	4.7	7
181	The Neuroscience of Glioblastoma. Molecular Pathology Library, 2021, , 247-259.	0.1	0
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183	Neurosonographic Approach to Malformations of Cortical Development. Donald School Journal of Ultrasound in Obstetrics and Gynecology, 2021, 15, 179-187.	0.3	0
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185	Wilms Tumor. , 2021, , 139-162.		0
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