

Heterozygous Mutations in Natriuretic Peptide Receptor Short Stature

Human Mutation

36, 474-481

DOI: [10.1002/humu.22773](https://doi.org/10.1002/humu.22773)

Citation Report

#	ARTICLE	IF	CITATIONS
1	Heterozygous NPR2 Mutations Cause Disproportionate Short Stature, Similar to Léri-Weill Dyschondrosteosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015, 100, E1133-E1142.	3.6	60
2	Short and tall stature: a new paradigm emerges. <i>Nature Reviews Endocrinology</i> , 2015, 11, 735-746.	9.6	212
3	Genomic insights into growth and its disorders. <i>Current Opinion in Endocrinology, Diabetes and Obesity</i> , 2016, 23, 51-56.	2.3	10
4	Reflections on the US Guidelines on Growth Hormone and Insulin-Like Growth Factor-I Treatment in Children and Adolescents. <i>Hormone Research in Paediatrics</i> , 2016, 86, 398-402.	1.8	5
5	Novel mutations in the transmembrane natriuretic peptide receptor NPR-B gene in four Indian families with acromesomelic dysplasia, type Maroteaux. <i>Journal of Genetics</i> , 2016, 95, 905-909.	0.7	7
6	Molecular Physiology of Membrane Guanylyl Cyclase Receptors. <i>Physiological Reviews</i> , 2016, 96, 751-804.	28.8	291
7	Genetic Techniques in the Evaluation of Short Stature. <i>Endocrinology and Metabolism Clinics of North America</i> , 2016, 45, 345-358.	3.2	6
8	A Track Record on SHOX: From Basic Research to Complex Models and Therapy. <i>Endocrine Reviews</i> , 2016, 37, 417-448.	20.1	87
9	Acromesomelic dysplasia, type maroteaux caused by novel loss-of-function mutations of the NPR2 gene: Three case reports. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 426-434.	1.2	28
10	MECHANISMS IN ENDOCRINOLOGY: Novel genetic causes of short stature. <i>European Journal of Endocrinology</i> , 2016, 174, R145-R173.	3.7	134
11	Catalytically Active Guanylyl Cyclase B Requires Endoplasmic Reticulum-mediated Glycosylation, and Mutations That Inhibit This Process Cause Dwarfism. <i>Journal of Biological Chemistry</i> , 2016, 291, 11385-11393.	3.4	19
12	Skeletal overgrowth-causing mutations mimic an allosterically activated conformation of guanylyl cyclase-B that is inhibited by 2,4,6-trinitrophenyl ATP. <i>Journal of Biological Chemistry</i> , 2017, 292, 10220-10229.	3.4	4
13	New Genetic Diagnoses of Short Stature Provide Insights into Local Regulation of Childhood Growth. <i>Hormone Research in Paediatrics</i> , 2017, 88, 22-37.	1.8	29
14	Identification of a novel heterozygous mutation of the Aggrecan gene in a family with idiopathic short stature and multiple intervertebral disc herniation. <i>Journal of Human Genetics</i> , 2017, 62, 717-721.	2.3	29
15	Long-term response to growth hormone therapy in a patient with short stature caused by a novel heterozygous mutation in NPR2. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2017, 30, 111-116.	0.9	14
16	Next generation sequencing-based mutation screening of 86 patients with idiopathic short stature. <i>Endocrine Journal</i> , 2017, 64, 947-954.	1.6	41
17	Novel genetic cause of idiopathic short stature. <i>Annals of Pediatric Endocrinology and Metabolism</i> , 2017, 22, 153-157.	2.3	24
18	Mutations in C-natriuretic peptide (NPPC): a novel cause of autosomal dominant short stature. <i>Genetics in Medicine</i> , 2018, 20, 91-97.	2.4	49

#	ARTICLE	IF	CITATIONS
19	Children Born Small for Gestational Age: Differential Diagnosis, Molecular Genetic Evaluation, and Implications. <i>Endocrine Reviews</i> , 2018, 39, 851-894.	20.1	122
20	Plasma C-Type Natriuretic Peptide: Emerging Applications in Disorders of Skeletal Growth. <i>Hormone Research in Paediatrics</i> , 2018, 90, 345-357.	1.8	15
21	Heterozygous NPR2 Mutation in Two Family Members with Short Stature and Skeletal Dysplasia. Case Reports in <i>Endocrinology</i> , 2018, 2018, 1-4.	0.4	7
22	PAPPA2 as a Therapeutic Modulator of IGF-I Bioavailability: in Vivo and in Vitro Evidence. <i>Journal of the Endocrine Society</i> , 2018, 2, 646-656.	0.2	19
23	Skeletal Dysplasias. , 2018, , 175-196.		1
24	C-type natriuretic peptide analog treatment of craniosynostosis in a Crouzon syndrome mouse model. <i>PLoS ONE</i> , 2018, 13, e0201492.	2.5	7
25	Natriuretic Peptides and Normal Body Fluid Regulation. , 2018, 8, 1211-1249.		30
26	Molecular and in silico analyses validates pathogenicity of homozygous mutations in the NPR2 gene underlying variable phenotypes of Acromesomelic dysplasia, type Maroteaux. <i>International Journal of Biochemistry and Cell Biology</i> , 2018, 102, 76-86.	2.8	14
27	Analysis of short-term treatment with the phosphodiesterase type 5 inhibitor tadalafil on long bone development in young rats. <i>American Journal of Physiology - Endocrinology and Metabolism</i> , 2018, 315, E446-E453.	3.5	6
28	Genetics of Growth Disorders – Which Patients Require Genetic Testing?. <i>Frontiers in Endocrinology</i> , 2019, 10, 602.	3.5	33
29	Towards a Rational and Efficient Diagnostic Approach in Children Referred for Growth Failure to the General Paediatrician. <i>Hormone Research in Paediatrics</i> , 2019, 91, 223-240.	1.8	37
30	Genetic regulation of linear growth. <i>Annals of Pediatric Endocrinology and Metabolism</i> , 2019, 24, 2-14.	2.3	10
31	Genetic causes of isolated short stature. <i>Archives of Endocrinology and Metabolism</i> , 2019, 63, 70-78.	0.6	24
32	Multigene Sequencing Analysis of Children Born Small for Gestational Age With Isolated Short Stature. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019, 104, 2023-2030.	3.6	55
33	Growth hormone therapy in children; research and practice – A review. <i>Growth Hormone and IGF Research</i> , 2019, 44, 20-32.	1.1	52
34	CREB activation in hypertrophic chondrocytes is involved in the skeletal overgrowth in epiphyseal chondrodysplasia Miura type caused by activating mutations of natriuretic peptide receptor B. <i>Human Molecular Genetics</i> , 2019, 28, 1183-1198.	2.9	8
35	Novel variants in natriuretic peptide receptor 2 in unrelated patients with acromesomelic dysplasia type Maroteaux. <i>European Journal of Medical Genetics</i> , 2019, 62, 103554.	1.3	12
36	Abnormal Body Size and Proportion. , 2019, , 81-143.		0

#	ARTICLE	IF	CITATIONS
37	Short Stature is Progressive in Patients with Heterozygous NPR2 Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 3190-3202.	3.6	15
38	Biallelic cGMP-dependent type II protein kinase gene (<i>PRKG2</i>) variants cause a novel acromesomic dysplasia. <i>Journal of Medical Genetics</i> , 2022, 59, 28-38.	3.2	10
39	Novel Mutations and Genes That Impact on Growth in Short Stature of Undefined Aetiology: The EPIGROW Study. <i>Journal of the Endocrine Society</i> , 2020, 4, bvaa105.	0.2	10
40	Tremor is a major feature of 9p13 deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2694-2698.	1.2	1
41	Male mice with elevated C-type natriuretic peptide-dependent guanylyl cyclase-B activity have increased osteoblasts, bone mass and bone strength. <i>Bone</i> , 2020, 135, 115320.	2.9	17
42	C-type natriuretic peptide stimulates osteoblastic proliferation and collagen-X expression but suppresses fibroblast growth factor-23 expression in vitro. <i>Pediatric Rheumatology</i> , 2020, 18, 46.	2.1	6
43	Role of <i>NPR2</i> mutation in idiopathic short stature: Identification of two novel mutations. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1146.	1.2	18
44	<i>NPR2</i> Variants Are Frequent among Children with Familiar Short Stature and Respond Well to Growth Hormone Therapy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, e746-e752.	3.6	26
45	An Activating Deletion Variant in the Submembrane Region of Natriuretic Peptide Receptor-B Causes Tall Stature. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2354-2366.	3.6	5
46	Clinical and Molecular Description of 16 Families With Heterozygous <i>IHH</i> Variants. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2654-2666.	3.6	6
47	Growth plate gene involvement and isolated short stature. <i>Endocrine</i> , 2021, 71, 28-34.	2.3	18
48	Clinical Characteristics of Short-Stature Patients With an <i>NPR2</i> Mutation and the Therapeutic Response to rhGH. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, 431-441.	3.6	15
49	Disorders of Childhood Growth. , 2021, , 299-356.		3
50	Prevention of guanylyl cyclase-B dephosphorylation rescues achondroplastic dwarfism. <i>JCI Insight</i> , 2021, 6, .	5.0	12
51	De novo c.2455C>T mutation of <i>NPR2</i> gene in a fetus with shortened long bones and a ventricular septal defect conceived by a mother with a fragile site at 16q22.1 and a father with a rare heterochromatic variant of chromosome 4 from Vietnam. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1637.	1.2	0
52	Growth failure: "idiopathic" only after a detailed diagnostic evaluation. <i>Endocrine Connections</i> , 2021, 10, R125-R138.	1.9	17
53	Identifying therapeutic drug targets using bidirectional effect genes. <i>Nature Communications</i> , 2021, 12, 2224.	12.8	11
54	Association Between Natriuretic Peptide Receptor 2 (<i>NPR2</i>) <i>RS208158047</i> Polymorphism and Fattening Performance of Young Bulls. <i>Annals of Animal Science</i> , 2022, 22, 109-119.	1.6	1

#	ARTICLE	IF	CITATIONS
55	Acromesomelic dysplasia-Maroteaux type, nine patients with two novel NPR2 variants. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 1115-1121.	0.9	0
56	<i>NPR2</i> gene variants in familial short stature: a single-center study. Journal of Pediatric Endocrinology and Metabolism, 2022, 35, 185-190.	0.9	4
57	Hormone resistance and short stature: A journey through the pathways of hormone signaling. Molecular and Cellular Endocrinology, 2021, 536, 111416.	3.2	1
60	Circulating osteocrin stimulates bone growth by limiting C-type natriuretic peptide clearance. Journal of Clinical Investigation, 2017, 127, 4136-4147.	8.2	43
61	A novel <i>NPR2</i> mutation (p.Arg388Gln) in a patient with acromesomelic dysplasia, type Maroteaux. Clinical Pediatric Endocrinology, 2020, 29, 99-103.	0.8	3
62	Rats deficient C-type natriuretic peptide suffer from impaired skeletal growth without early death. PLoS ONE, 2018, 13, e0194812.	2.5	14
63	GHR gene transcript heterogeneity may explain phenotypic variability in GHR pseudoexon (6 ^h) patients. Endocrine Connections, 2020, 9, 211-222.	1.9	4
64	Achieving Optimal Short- and Long-term Responses to Paediatric Growth Hormone Therapy. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 329-340.	0.9	16
65	Application of Genomics to the Study of Human Growth Disorders. Translational Bioinformatics, 2016, , 363-384.	0.0	0
66	Natriuretic Peptide Receptor Type B (NPRB). , 2017, , 1-5.		0
67	Mutant mouse tells the function of CNP/NPR-B in the smooth muscle. Journal of Animal Genetics, 2017, 45, 9-18.	1.0	0
68	Natriuretic Peptide Receptor Type B (NPRB). , 2018, , 3351-3355.		0
69	Expanding horizons of achondroplasia treatment: current options and future developments. Osteoarthritis and Cartilage, 2021, , .	1.3	4
70	Short Stature in Children Born Small for Gestational Age. , 2022, , 1124-1135.		0
71	Broadening the spectrum of loss-of-function variants in NPR-C-related extreme tall stature. Journal of the Endocrine Society, 2022, 6, bvac019.	0.2	2
72	Analysis of rare genetic variation underlying cardiometabolic diseases and traits among 200,000 individuals in the UK Biobank. Nature Genetics, 2022, 54, 240-250.	21.4	68
73	Balanced assessment of growth disorders using clinical, endocrinological, and genetic approaches. Annals of Pediatric Endocrinology and Metabolism, 2021, 26, 218-226.	2.3	11
74	Should Skeletal Maturation Be Manipulated for Extra Height Gain?. Frontiers in Endocrinology, 2021, 12, 812196.	3.5	9

#	ARTICLE	IF	CITATIONS
75	Heterozygous NPR2 Variants in Idiopathic Short Stature. <i>Genes</i> , 2022, 13, 1065.	2.4	4
76	Circulating exosomal circRNA_0063476 impairs expression of markers of bone growth via the miR-518c-3p/DDX6 axis in ISS. <i>Endocrinology</i> , 0, , .	2.8	1
77	Identification of Diagnostic Variants in <i>FGFR2</i> and <i>NPR2</i> Genes in a Chinese Family Affected by Crouzon Syndrome and Acromesomelic Dysplasia, Type Maroteaux. <i>DNA and Cell Biology</i> , 0, , .	1.9	0
78	International Consensus Guideline on Small for Gestational Age: Etiology and Management From Infancy to Early Adulthood. <i>Endocrine Reviews</i> , 2023, 44, 539-565.	20.1	16
79	Acromesomelic Dysplasia I, Maroteaux Type (AMD1). , 2023, , 1-4.		0
80	Assessment of Nutritional Status in the Diagnostic Evaluation of the Child with Growth Failure. <i>Hormone Research in Paediatrics</i> , 2024, 97, 11-21.	1.8	0
81	Clinical and genetic evaluation of children with short stature of unknown origin. <i>BMC Medical Genomics</i> , 2023, 16, .	1.5	2
82	Two new patients with acromesomelic dysplasia, PRKG2 type identification and characterization of the first missense variant. <i>European Journal of Human Genetics</i> , 0, , .	2.8	0
83	Exploring the Genetic Causes for Postnatal Growth Failure in Children Born Non-Small for Gestational Age. <i>Journal of Clinical Medicine</i> , 2023, 12, 6508.	2.4	0
84	Identification of novel genes including NAV2 associated with isolated tall stature. <i>Frontiers in Endocrinology</i> , 0, 14, .	3.5	0
85	Pubertal growth in children born small for gestational age (SGA) with persistent short stature (SGA-SS). Growth hormone treatment outcomes from data of the Czech countywide REPAR database. <i>Cesko-Slovenska Pediatrie</i> , 2024, 78, 30-36.	0.2	0
86	C-Type Natriuretic Peptide Analogs: Current and Future Therapeutic Applications. <i>Hormone Research in Paediatrics</i> , 0, , 1-1.	1.8	0