

# Bardet-Biedl syndrome

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Patient with Bardet-Biedl syndrome presenting with nystagmus at fifteen months of age. <i>Journal of AAPOS</i> , 2001, 5, 262-264.	0.2	5
2	Genetic Disorders Among Arab Populations. , 2010, , .		45
3	Spectrum of Clinical Diseases Caused By Disorders of Primary Cilia. <i>Proceedings of the American Thoracic Society</i> , 2011, 8, 444-450.	3.5	97
4	The <i>L. eishmania major</i> BBSome subunit BBS 1 is essential for parasite virulence in the mammalian host. <i>Molecular Microbiology</i> , 2013, 90, 597-611.	1.2	15
5	Further cause for concern in childhood vision impairment. <i>Clinical and Experimental Ophthalmology</i> , 2013, 41, 727-728.	1.3	1
6	Crystal structure of the small GTPase <i>Arl6/BBS3</i> from <i>Trypanosoma brucei</i> . <i>Protein Science</i> , 2013, 22, 196-203.	3.1	4
7	Bardet-Biedl syndrome presenting as dilated cardiomyopathy. <i>Journal of Indian College of Cardiology</i> , 2013, 3, 134-138.	0.1	1
8	Delirious Mania Associated with Bardet-Biedl Syndrome, an Inherited Ciliopathy. <i>Psychosomatics</i> , 2013, 54, 484-487.	2.5	0
9	Rab and Arf Proteins in Genetic Diseases. <i>Traffic</i> , 2013, 14, 871-885.	1.3	48
10	Gene Therapy for Blindness. <i>Annual Review of Neuroscience</i> , 2013, 36, 467-488.	5.0	124
11	Cystic Diseases of the Kidney. , 2013, , 1-33.		0
12	Delineation of the Key Aspects in the Regulation of Epithelial Monolayer Formation. <i>Molecular and Cellular Biology</i> , 2013, 33, 2535-2550.	1.1	71
13	Evolution of modular intraflagellar transport from a coatomer-like progenitor. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 6943-6948.	3.3	144
14	Siblings with Bardet Beidl Syndrome. <i>Journal of Nepal Paediatric Society</i> , 2013, 33, 236-238.	0.1	0
15	The Case   Hexadactyly, blindness, obesity, and end-stage renal disease. <i>Kidney International</i> , 2013, 84, 1291-1292.	2.6	2
16	How to approach endocrine assessment in severe obesity?. <i>Clinical Endocrinology</i> , 2013, 79, 163-167.	1.2	14
17	Leptin signaling defects in a mouse model of Prader-Willi syndrome. <i>Rare Diseases (Austin, Tex )</i> , 2013, 1, e24421.	1.8	8
18	A Complex of BBS1 and NPHP7 Is Required for Cilia Motility in Zebrafish. <i>PLoS ONE</i> , 2013, 8, e72549.	1.1	21

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19	The Genetics of Reading Disabilities: From Phenotypes to Candidate Genes. <i>Frontiers in Psychology</i> , 2013, 3, 601.	1.1	51
20	Comprehensive Molecular Diagnosis of Bardet-Biedl Syndrome by High-Throughput Targeted Exome Sequencing. <i>PLoS ONE</i> , 2014, 9, e90599.	1.1	42
21	Knockdown of the BBS10 Gene Product Affects Apical Targeting of AQP2 in Renal Cells: A Possible Explanation for the Polyuria Associated with Bardet-Biedl Syndrome. <i>Journal of Genetic Syndromes &amp; Gene Therapy</i> , 2014, 05, .	0.2	5
22	Report of four cases of Bardet-Biedl syndrome. <i>Jornal Brasileiro De Nefrologia: Orgao Oficial De Sociedades Brasileira E Latino-Americana De Nefrologia</i> , 2014, 36, 250-253.	0.4	2
23	Bardet-Biedl syndrome and a large concha bullosa pyocele. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014, 78, 2316-2318.	0.4	1
25	Cilia and Diseases. <i>BioScience</i> , 2014, 64, 1126-1137.	2.2	167
26	Primary cilia enhance kisspeptin receptor signaling on gonadotropin-releasing hormone neurons. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2014, 111, 10335-10340.	3.3	81
27	IFT27, encoding a small GTPase component of IFT particles, is mutated in a consanguineous family with Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2014, 23, 3307-3315.	1.4	134
28	Deciphering intrafamilial phenotypic variability by exome sequencing in a Bardet-Biedl family. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2014, 2, 124-133.	0.6	13
29	Essential Role of the Chaperonin CCT in Rod Outer Segment Biogenesis. , 2014, 55, 3775.		12
30	Overview of Bardet-Biedl syndrome in Spain: identification of novel mutations in <i>BBS1</i> , <i>BBS10</i> and <i>BBS12</i> genes. <i>Clinical Genetics</i> , 2014, 86, 601-602.	1.0	20
31	Bardet-Biedl Syndrome with Urogenital Sinus Presenting with Acute Renal Failure in a Neonate. <i>Indian Journal of Pediatrics</i> , 2014, 81, 719-721.	0.3	1
32	Functional modelling of a novel mutation in BBS5. <i>Cilia</i> , 2014, 3, 3.	1.8	13
33	Cell-free DNA testing: An aid to prenatal sonographic diagnosis. <i>Best Practice and Research in Clinical Obstetrics and Gynaecology</i> , 2014, 28, 453-466.	1.4	7
34	The neuropathology of obesity: insights from human disease. <i>Acta Neuropathologica</i> , 2014, 127, 3-28.	3.9	64
35	Contribution of Mutation Load to the Intrafamilial Genetic Heterogeneity in a Large Cohort of Spanish Retinal Dystrophies Families. , 2014, 55, 7562.		11
36	Small organelle, big responsibility: the role of centrosomes in development and disease. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2014, 369, 20130468.	1.8	128
37	A Novel Test for Recessive Contributions to Complex Diseases Implicates Bardet-Biedl Syndrome Gene BBS10 in Idiopathic Type 2 Diabetes and Obesity. <i>American Journal of Human Genetics</i> , 2014, 95, 509-520.	2.6	29

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38	Mutation profile of BBS genes in Iranian patients with Bardet-Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. <i>Journal of Human Genetics</i> , 2014, 59, 368-375.	1.1	33
39	Role of cilia in normal pancreas function and in diseased states. <i>Birth Defects Research Part C: Embryo Today Reviews</i> , 2014, 102, 126-138.	3.6	20
40	The genetics of human obesity. <i>Translational Research</i> , 2014, 164, 293-301.	2.2	70
41	Intravenous Injections in Neonatal Mice. <i>Journal of Visualized Experiments</i> , 2014, , e52037.	0.2	41
42	Update on the Genetics of Bardet-Biedl Syndrome. <i>Molecular Syndromology</i> , 2014, 5, 51-56.	0.3	98
43	General and Stomatologic Aspects of Bardet-Biedl Syndrome. <i>Journal of Craniofacial Surgery</i> , 2014, 25, e575-e578.	0.3	2
44	Nasal Nitric Oxide in Patients With Inherited Retinal Dystrophies. <i>Journal of Investigative Medicine</i> , 2015, 63, 554-557.	0.7	3
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46	Comparing the Bbs10 complete knockout phenotype with a specific renal epithelial knockout one highlights the link between renal defects and systemic inactivation in mice. <i>Cilia</i> , 2015, 4, 10.	1.8	29
47	Identification of Two Cases of Ciliopathy-Associated Diabetes and Their Mutation Analysis Using Whole Exome Sequencing. <i>Diabetes and Metabolism Journal</i> , 2015, 39, 439.	1.8	6
48	Advances in the understanding of the BBSome complex structure and function. <i>Research and Reports in Biology</i> , 0, , 191.	0.2	5
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50	Cilia Ift protein and motor -related bone diseases and mouse models. <i>Frontiers in Bioscience - Landmark</i> , 2015, 20, 515-555.	3.0	29
51	Molecular Pathways Underlying Projection Neuron Production and Migration during Cerebral Cortical Development. <i>Frontiers in Neuroscience</i> , 2015, 9, 447.	1.4	79
52	Characterization of Courtesy Stigma Perceived by Parents of Overweight Children with Bardet-Biedl Syndrome. <i>PLoS ONE</i> , 2015, 10, e0140705.	1.1	33
53	Two Brothers with Bardet-Biedl Syndrome Presenting with Chronic Renal Failure. <i>Case Reports in Nephrology</i> , 2015, 2015, 1-5.	0.2	3
54	Type 2 diabetes presenting with hyperglycaemic hyperosmolar state in an adolescent renal transplant patient. <i>BMJ Case Reports</i> , 2015, 2015, bcr2014207124-bcr2014207124.	0.2	2
55	A Case of Hydrometrocolpos and Polydactyly. <i>Clinical Medicine Insights Pediatrics</i> , 2015, 9, CMPed.S20787.	0.7	9

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56	Contrasting genetic architectures in different mouse reference populations used for studying complex traits. <i>Genome Research</i> , 2015, 25, 775-791.	2.4	56
57	Syndromic ciliopathies: From single gene to multi gene analysis by SNP arrays and next generation sequencing. <i>Molecular and Cellular Probes</i> , 2015, 29, 299-307.	0.9	27
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59	Neural Regulatory Pathways of Feeding and Fat in <i>Caenorhabditis elegans</i> . <i>Annual Review of Genetics</i> , 2015, 49, 413-438.	3.2	39
60	Algorithm for the molecular analysis of Bardet-Biedl syndrome in Spain. <i>Medicina Clínica (English)</i> 130(10):1071-1072	0.1	2
61	Ciliary proteins <i>Bbs8</i> and <i>Ift20</i> promote planar cell polarity in the cochlea. <i>Development (Cambridge)</i> , 2015, 142, 555-566.	1.2	63
62	Cystic Kidney Disease: A Primer. <i>Advances in Chronic Kidney Disease</i> , 2015, 22, 297-305.	0.6	34
63	Genome-wide association study of acute kidney injury after coronary bypass graft surgery identifies susceptibility loci. <i>Kidney International</i> , 2015, 88, 823-832.	2.6	42
64	Bardet-Biedl syndrome: Is it only cilia dysfunction?. <i>FEBS Letters</i> , 2015, 589, 3479-3491.	1.3	77
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66	Exploring genotype-phenotype relationships in Bardet-Biedl syndrome families. <i>Journal of Medical Genetics</i> , 2015, 52, 503-513.	1.5	42
67	Bardet Biedl Syndrome. <i>Chest</i> , 2015, 147, 764-770.	0.4	24
68	Evaluation of Zebrafish Kidney Function Using a Fluorescent Clearance Assay. <i>Journal of Visualized Experiments</i> , 2015, , e52540.	0.2	25
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72	Ciliary disturbances in syndromal and non-syndromal obesity. <i>Journal of Pediatric Genetics</i> , 2015, 03, 079-088.	0.3	2
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76	Clinical Characteristics and Current Therapies for Inherited Retinal Degenerations. <i>Cold Spring Harbor Perspectives in Medicine</i> , 2015, 5, a017111-a017111.	2.9	171
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78	Function and regulation of primary cilia and intraflagellar transport proteins in the skeleton. <i>Annals of the New York Academy of Sciences</i> , 2015, 1335, 78-99.	1.8	86
79	Mutation spectrum in <scp>BBS</scp> genes guided by homozygosity mapping in an Indian cohort. <i>Clinical Genetics</i> , 2015, 87, 161-166.	1.0	28
80	Genetic predictors of cardiovascular morbidity in Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2015, 87, 343-349.	1.0	27
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83	Bardet Biedl syndrome in South Africa: A single founder mutation. <i>South African Medical Journal</i> , 2016, 106, 72.	0.2	6
84	Anesthetic considerations for patients with Bardet-Biedl syndrome: a case series and review of the literature. <i>Paediatric Anaesthesia</i> , 2016, 26, 429-437.	0.6	5
85	Genetics of human Bardet-Biedl syndrome, an updates. <i>Clinical Genetics</i> , 2016, 90, 3-15.	1.0	128
86	TMEM107 Is a Critical Regulator of Ciliary Protein Composition and Is Mutated in Orofaciodigital Syndrome. <i>Human Mutation</i> , 2016, 37, 155-159.	1.1	18
87	A compendium of human genes regulating feeding behavior and body weight, its functional characterization and identification of GWAS genes involved in brain-specific PPI network. <i>BMC Genetics</i> , 2016, 17, 158.	2.7	15
88	Cilia and Flagella. , 2016, , 660-676.		1
89	Mutational and clinical analysis of the ENG gene in patients with pulmonary arterial hypertension. <i>BMC Genetics</i> , 2016, 17, 72.	2.7	21
90	Signaling Pathways Involved in Mammalian Sex Determination and Gonad Development. <i>Sexual Development</i> , 2015, 9, 297-315.	1.1	84
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95	Mutations in the polyglutamylase gene <i>TLL5</i> , expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. <i>Human Molecular Genetics</i> , 2016, 25, ddw282.	1.4	27
96	Bardet-Biedl Syndrome. <i>Molecular Syndromology</i> , 2016, 7, 62-71.	0.3	103
97	Overlap of abnormal photoreceptor development and progressive degeneration in Leber congenital amaurosis caused by <i>NPHP5</i> mutation. <i>Human Molecular Genetics</i> , 2016, 25, 4211-4226.	1.4	35
98	Tumor Necrosis Factor $\alpha$ Impairs Kisspeptin Signaling in Human Gonadotropin-Releasing Hormone Primary Neurons. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, jc.2016-2115.	1.8	47
99	<i>The brain needs interleukin-6 (IL-6) to maintain a "healthy" energy balance</i> . Focus on "IL-6 ameliorates defective leptin sensitivity in DIO ventromedial hypothalamic nucleus neurons" <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , 2016, 311, R989-R991.	0.9	6
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103	Investigating Connections between Metabolism, Longevity, and Behavior in <i>Caenorhabditis elegans</i> . <i>Trends in Endocrinology and Metabolism</i> , 2016, 27, 586-596.	3.1	39
104	Aberrant protein trafficking in retinal degenerations: The initial phase of retinal remodeling. <i>Experimental Eye Research</i> , 2016, 150, 71-80.	1.2	26
105	<i>C8orf37</i> is mutated in Bardet-Biedl syndrome and constitutes a locus allelic to non-syndromic retinal dystrophies. <i>Ophthalmic Genetics</i> , 2016, 37, 290-293.	0.5	29
106	Non-essential role for cilia in coordinating precise alignment of lens fibres. <i>Mechanisms of Development</i> , 2016, 139, 10-17.	1.7	13
107	Differential effects on $\beta$ -cell mass by disruption of Bardet-Biedl syndrome or Alstrom syndrome genes. <i>Human Molecular Genetics</i> , 2016, 25, 57-68.	1.4	35
108	Bardet-Biedl syndrome: A model for translational research in rare diseases. <i>European Journal of Molecular and Clinical Medicine</i> , 2017, 2, 102.	0.5	5
110	Cilia and Obesity. <i>Cold Spring Harbor Perspectives in Biology</i> , 2017, 9, a028217.	2.3	84
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112	The CEP19-RABL2 GTPase Complex Binds IFT-B to Initiate Intraflagellar Transport at the Ciliary Base. <i>Developmental Cell</i> , 2017, 42, 22-36.e12.	3.1	105
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116	Role of Primary Cilia in Odontogenesis. <i>Journal of Dental Research</i> , 2017, 96, 965-974.	2.5	39
117	A systematic review of genetic syndromes with obesity. <i>Obesity Reviews</i> , 2017, 18, 603-634.	3.1	138
118	Renal cystic disease and associated ciliopathies. <i>Current Opinion in Obstetrics and Gynecology</i> , 2017, 29, 85-94.	0.9	31
119	Ciliopathies: Genetics in Pediatric Medicine. <i>Journal of Pediatric Genetics</i> , 2017, 06, 018-029.	0.3	46
120	A novel BBS10 mutation identified in a patient with Bardet-Biedl syndrome with a violent emotional outbreak. <i>Human Genome Variation</i> , 2017, 4, 17033.	0.4	4
121	The genetic underpinnings of body fat distribution. <i>Expert Review of Endocrinology and Metabolism</i> , 2017, 12, 417-427.	1.2	3
122	The Kidney in Bardet-Biedl Syndrome: Possible Pathogenesis of Urine Concentrating Defect. <i>Kidney Diseases (Basel, Switzerland)</i> , 2017, 3, 57-65.	1.2	14
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127	Oral healthcare management in Bardet Biedl syndrome. <i>Special Care in Dentistry</i> , 2017, 37, 47-50.	0.4	3
128	Risk Factors for Severe Renal Disease in Bardet-Biedl Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 963-970.	3.0	69
129	Six-transmembrane epithelial antigens of the prostate comprise a novel inflammatory nexus in patients with pustular skin disorders. <i>Journal of Allergy and Clinical Immunology</i> , 2017, 139, 1217-1227.	1.5	38



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131	New Thoughts on Pediatric Genetic Obesity: Pathogenesis, Clinical Characteristics and Treatment Approach. , 0, , .		0
132	Ophthalmic manifestations of endocrine disorders”endocrinology and the eye. Translational Pediatrics, 2017, 6, 286-299.	0.5	9
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134	Beneficial Outcomes of Sleeve Gastrectomy in a Morbidly Obese Patient With Bardet-Biedl Syndrome. Journal of the Endocrine Society, 2017, 1, 317-322.	0.1	15
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137	The dilemma of diagnostic testing for Prader-Willi syndrome. Translational Pediatrics, 2017, 5, 46-56.	0.5	23
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147	The Genetic Basis of Delayed Puberty. Neuroendocrinology, 2018, 106, 283-291.	1.2	40

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149	Hearing loss and renal syndromes. <i>Pediatric Nephrology</i> , 2018, 33, 1671-1683.	0.9	12
150	Homozygous mutation in <i>CEP190</i> gene mutated in morbid obesity, in Bardet-Biedl syndrome with predominant postaxial polydactyly. <i>Journal of Medical Genetics</i> , 2018, 55, 189-197.	1.5	25
151	OligoPVP: Phenotype-driven analysis of individual genomic information to prioritize oligogenic disease variants. <i>Scientific Reports</i> , 2018, 8, 14681.	1.6	8
152	Revisiting the evolutionary origins of obesity: lazy versus peppy-thrifty genotype hypothesis. <i>Obesity Reviews</i> , 2018, 19, 1525-1543.	3.1	19
153	Variante patogénica homocigótica del gen BBS10 en un paciente con síndrome de Bardet-Biedl. <i>Biomedica</i> , 2018, 38, 308-320.	0.3	0
155	Not quite type 1 or type 2, what now? Review of monogenic, mitochondrial, and syndromic diabetes. <i>Reviews in Endocrine and Metabolic Disorders</i> , 2018, 19, 35-52.	2.6	25
156	Ciliary Defects in a Mouse Model of Bardet-Biedl Syndrome are Selectively Pronounced in Brain Regions Involved in Cardiovascular Regulation. <i>Neuroscience and Behavioral Physiology</i> , 2018, 48, 28-38.	0.2	0
157	Managing Bardet-Biedl Syndrome Now and in the Future. <i>Frontiers in Pediatrics</i> , 2018, 6, 23.	0.9	136
158	Network for Early Onset Cystic Kidney Diseases: A Comprehensive Multidisciplinary Approach to Hereditary Cystic Kidney Diseases in Childhood. <i>Frontiers in Pediatrics</i> , 2018, 6, 24.	0.9	19
159	Severe skeletal abnormalities caused by defects in retrograde intraflagellar transport dyneins. , 2018, , 356-401.		7
160	The Endocrine and Metabolic Characteristics of a Large Bardet-Biedl Syndrome Clinic Population. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 1834-1841.	1.8	58
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