

# A Novel Homozygous Nonsense Mutation in *CABP4* <br>Synaptic Disorder

DOI: 10.1167/iops.08-2553

Citation Report

#	ARTICLE	IF	CITATIONS
1	Genotyping Microarray for CSNB-Associated Genes. , 2009, 50, 5919.		41
2	Visually impaired children: â€œcoming to better termsâ€ Documenta Ophthalmologica, 2009, 119, 1-7.	1.0	6
3	Bioinformatic analysis of CaBP/calneuron proteins reveals a family of highly conserved vertebrate Ca <sup>2+</sup> -binding proteins. BMC Research Notes, 2010, 3, 118.	0.6	25
4	Homozygosity Mapping in Patients with Coneâ€“Rod Dystrophy: Novel Mutations and Clinical Characterizations. , 2010, 51, 5943.		92
5	The Diversity of Calcium Sensor Proteins in the Regulation of Neuronal Function. Cold Spring Harbor Perspectives in Biology, 2010, 2, a004085-a004085.	2.3	83
6	Homozygosity mapping: One more tool in the clinical geneticist's toolbox. Genetics in Medicine, 2010, 12, 236-239.	1.1	107
7	Insight into the Role of Ca <sup>2+</sup> -Binding Protein 5 in Vesicle Exocytosis. , 2011, 52, 9131.		12
8	An extended 15ÂHz ERG protocol (1): the contributions of primary and secondary rod pathways and the cone pathway. Documenta Ophthalmologica, 2011, 123, 149-159.	1.0	10
9	An extended 15ÂHz ERG protocol (2): data of normal subjects and patients with achromatopsia, CSNB1, and CSNB2. Documenta Ophthalmologica, 2011, 123, 161-172.	1.0	7
10	Animal Models of Retinal Disease. Progress in Molecular Biology and Translational Science, 2011, 100, 211-286.	0.9	89
11	Complex Regulation of Voltage-dependent Activation and Inactivation Properties of Retinal Voltage-gated Cav1.4 L-type Ca <sup>2+</sup> Channels by Ca <sup>2+</sup> -binding Protein 4 (CaBP4)*. Journal of Biological Chemistry, 2012, 287, 36312-36321.	1.6	43
12	A Mutation in CABP2 , Expressed in Cochlear Hair Cells, Causes Autosomal-Recessive Hearing Impairment. American Journal of Human Genetics, 2012, 91, 636-645.	2.6	96
13	A vertex similarity-based framework to discover and rank orphan disease-related genes. BMC Systems Biology, 2012, 6, S8.	3.0	18
14	Mutations inRD3Are Associated with an Extremely Rare and Severe Form of Early Onset Retinal Dystrophy. , 2012, 53, 3463.		24
15	Evolution and functional diversity of the Calcium Binding Proteins (CaBPs). Frontiers in Molecular Neuroscience, 2012, 5, 9.	1.4	37
16	Retinal characteristics of the congenital disorder of glycosylation PMM2â€“CDG. Journal of Inherited Metabolic Disease, 2013, 36, 1039-1047.	1.7	16
17	Genotype and Phenotype of 101 Dutch Patients with Congenital Stationary Night Blindness. Ophthalmology, 2013, 120, 2072-2081.	2.5	95
18	Dysregulation of Ca <sup>v</sup> 1.4 channels disrupts the maturation of photoreceptor synaptic ribbons in congenital stationary night blindness type 2. Channels, 2013, 7, 514-523.	1.5	87

#	ARTICLE	IF	CITATIONS
19	Childhood retinal dystrophies: what's in a name?. <i>British Journal of Ophthalmology</i> , 2013, 97, 247-247.	2.1	4
20	Clinical characterisation of the <i>CABP4</i> -related retinal phenotype. <i>British Journal of Ophthalmology</i> , 2013, 97, 262-265.	2.1	28
21	Protein Phosphatase 2A Dephosphorylates CaBP4 and Regulates CaBP4 Function. , 2013, 54, 1214.		9
22	Assessment of Night Vision Problems in Patients with Congenital Stationary Night Blindness. <i>PLoS ONE</i> , 2013, 8, e62927.	1.1	34
23	Structural Insights into Activation of the Retinal L-type Ca <sup>2+</sup> Channel (Cav1.4) by Ca <sup>2+</sup> -binding Protein 4 (CaBP4). <i>Journal of Biological Chemistry</i> , 2014, 289, 31262-31273.	1.6	12
24	Anterior Cerebral Circulation Infarction and Retinal Ganglion Cell Degeneration. <i>Ophthalmology</i> , 2014, 121, e15-e16.	2.5	2
25	Mouse b-wave mutants. <i>Documenta Ophthalmologica</i> , 2014, 128, 77-89.	1.0	43
26	Vertebrate Photoreceptors. , 2014, , .		7
27	More Than a Pore: Ion Channel Signaling Complexes. <i>Journal of Neuroscience</i> , 2014, 34, 15159-15169.	1.7	62
28	Author reply. <i>Ophthalmology</i> , 2014, 121, e14-e15.	2.5	1
29	Localization and expression of CaBP1/caldendrin in the mouse brain. <i>Neuroscience</i> , 2014, 268, 33-47.	1.1	34
30	CABP4 Mutations Do Not Cause Congenital Stationary Night Blindness. <i>Ophthalmology</i> , 2014, 121, e15.	2.5	16
31	A COLQ Missense Mutation in Sphynx and Devon Rex Cats with Congenital Myasthenic Syndrome. <i>PLoS ONE</i> , 2015, 10, e0137019.	1.1	5
32	Congenital stationary night blindness: An analysis and update of genotype-phenotype correlations and pathogenic mechanisms. <i>Progress in Retinal and Eye Research</i> , 2015, 45, 58-110.	7.3	269
33	Sense and specificity in neuronal calcium signalling. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2015, 1853, 1921-1932.	1.9	48
34	Clinical Characteristics, Mutation Spectrum, and Prevalence of Å...land Eye Disease/Incomplete Congenital Stationary Night Blindness in Denmark. , 2016, 57, 6861.		25
35	Decalmodulation of Ca <sub>v</sub> 1 channels by CaBPs. <i>Channels</i> , 2016, 10, 33-37.	1.5	20
36	Leber congenital amaurosis, from darkness to light: An ode to Irene Maumenee. <i>Ophthalmic Genetics</i> , 2017, 38, 7-15.	0.5	28

#	ARTICLE	IF	CITATIONS
37	Coordinating the uncoordinated: UNC119 trafficking in cilia. <i>European Journal of Cell Biology</i> , 2017, 96, 643-652.	1.6	7
38	Development of Refractive Errors—What Can We Learn From Inherited Retinal Dystrophies?. <i>American Journal of Ophthalmology</i> , 2017, 182, 81-89.	1.7	61
39	Multimodal imaging in <i>CABP4</i> -related retinopathy. <i>Ophthalmic Genetics</i> , 2017, 38, 459-464.	0.5	10
40	Mutation screening of the <i>LRR13</i> , <i>CABP4</i> , and <i>GPR179</i> genes in Chinese patients with Schubert-Bornschein congenital stationary night blindness. <i>Ophthalmic Genetics</i> , 2017, 38, 206-210.	0.5	2
41	Retinal findings in a patient of French ancestry with <i>CABP4</i> -related retinal disease. <i>Documenta Ophthalmologica</i> , 2018, 136, 135-143.	1.0	4
42	CRISPR-Trap: a clean approach for the generation of gene knockouts and gene replacements in human cells. <i>Molecular Biology of the Cell</i> , 2018, 29, 75-83.	0.9	37
43	Genome-wide association meta-analysis highlights light-induced signaling as a driver for refractive error. <i>Nature Genetics</i> , 2018, 50, 834-848.	9.4	239
44	Channeling Vision: $Ca^{V1.4}$ —A Critical Link in Retinal Signal Transmission. <i>BioMed Research International</i> , 2018, 2018, 1-14.	0.9	30
45	Voltage-Gated Calcium Channels: Key Players in Sensory Coding in the Retina and the Inner Ear. <i>Physiological Reviews</i> , 2018, 98, 2063-2096.	13.1	79
46	Cone-rod homeobox <i>CRX</i> controls presynaptic active zone formation in photoreceptors of mammalian retina. <i>Human Molecular Genetics</i> , 2018, 27, 3555-3567.	1.4	17
47	Inferring novel genes related to colorectal cancer via random walk with restart algorithm. <i>Gene Therapy</i> , 2019, 26, 373-385.	2.3	3
48	Extracting the ON and OFF contributions to the full-field photopic flash electroretinogram using summed growth curves. <i>Experimental Eye Research</i> , 2019, 189, 107827.	1.2	6
49	Voltage- and calcium-gated ion channels of neurons in the vertebrate retina. <i>Progress in Retinal and Eye Research</i> , 2019, 72, 100760.	7.3	56
50	Calcium Sensors in Neuronal Function and Dysfunction. <i>Cold Spring Harbor Perspectives in Biology</i> , 2019, 11, a035154.	2.3	65
51	ROP18-Mediated Transcriptional Reprogramming of HEK293T Cell Reveals New Roles of ROP18 in the Interplay Between <i>Toxoplasma gondii</i> and the Host Cell. <i>Frontiers in Cellular and Infection Microbiology</i> , 2020, 10, 586946.	1.8	6
52	Clinical and Genetic Heterogeneity in Six Tunisian Families With Horizontal Gaze Palsy With Progressive Scoliosis: A Retrospective Study of 13 Cases. <i>Frontiers in Pediatrics</i> , 2020, 8, 172.	0.9	5
53	Large Animal Models of Inherited Retinal Degenerations: A Review. <i>Cells</i> , 2020, 9, 882.	1.8	47
54	The X-linked retinopathies: Physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. <i>Progress in Retinal and Eye Research</i> , 2021, 82, 100898.	7.3	65

#	ARTICLE	IF	CITATIONS
55	Negative electroretinograms: genetic and acquired causes, diagnostic approaches and physiological insights. <i>Eye</i> , 2021, 35, 2419-2437.	1.1	22
56	Congenital Stationary Night Blindness: Mutation Update and Clinical Variability. <i>Advances in Experimental Medicine and Biology</i> , 2012, 723, 371-379.	0.8	22
57	Exome sequencing identified a novel missense mutation c.464G>A (p.G155D) in Ca <sup>2+</sup> -binding protein 4 ( <i>CABP4</i> ) in a Chinese pedigree with autosomal dominant nocturnal frontal lobe epilepsy. <i>Oncotarget</i> , 2017, 8, 78940-78947.	0.8	17
58	Expression of CaBP transcripts in retinal bipolar cells of developing and adult zebrafish. <i>Matters</i> , 0, , .	1.0	4
59	Voltage-Gated Ca <sup>v</sup> 1 Channels in Disorders of Vision and Hearing. <i>Current Molecular Pharmacology</i> , 2015, 8, 143-148.	0.7	27
60	CaBP4. <i>The AFCS-nature Molecule Pages</i> , 0, , .	0.2	0
61	Molecular Mechanisms of Photoreceptor Synaptic Transmission. , 2014, , 167-198.		0
62	In silico structural and functional analysis of human calcium binding protein-5 (CaBP5). <i>International Journal of Pharma and Bio Sciences</i> , 2017, 8, .	0.1	0
63	Congenital Non-Degenerative Retinal Diseases. , 2019, , 37-50.		0
64	A null mutation in CABP4 causes Leber's congenital amaurosis-like phenotype. <i>Molecular Vision</i> , 2010, 16, 207-12.	1.1	28
65	A novel p.Gly603Arg mutation in CACNA1F causes Å...land island eye disease and incomplete congenital stationary night blindness phenotypes in a family. <i>Molecular Vision</i> , 2011, 17, 3262-70.	1.1	18
73	Ribbon Synapses and Retinal Disease: Review. <i>International Journal of Molecular Sciences</i> , 2023, 24, 5090.	1.8	2
75	Unusual OCT findings in a patient with CABP4-associated cone-rod synaptic disorder. <i>Documenta Ophthalmologica</i> , 2024, 148, 115-120.	1.0	0