

# Mutations in PNKP Cause Recessive Ataxia with Oculor

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Mutation in <i>PNKP</i> presenting initially as axonal Charcot-Marie-Tooth disease. <i>Neurology: Genetics</i> , 2015, 1, e30.	0.9	28
2	The Response to Oxidative DNA Damage in Neurons: Mechanisms and Disease. <i>Neural Plasticity</i> , 2016, 2016, 1-14.	1.0	56
3	Microcephalic primordial dwarfism in an Emirati patient with <i>PNKP</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2127-2132.	0.7	6
4	Overlapping roles for PARP1 and PARP2 in the recruitment of endogenous XRCC1 and PNKP into oxidized chromatin. <i>Nucleic Acids Research</i> , 2017, 45, gkw1246.	6.5	118
5	Expanding the ataxia with oculomotor apraxia type 4 phenotype. <i>Neurology: Genetics</i> , 2016, 2, e49.	0.9	37
6	DNA repair pathways underlie a common genetic mechanism modulating onset in polyglutamine diseases. <i>Annals of Neurology</i> , 2016, 79, 983-990.	2.8	183
7	Catastrophic cliffs: a partial suggestion for selective vulnerability in neurodegenerative diseases. <i>Biochemical Society Transactions</i> , 2016, 44, 659-661.	1.6	12
8	Hot topic: PNKP mutations cause ataxia with oculomotor apraxia type 4. <i>Movement Disorders</i> , 2016, 31, 500-500.	2.2	0
9	PNKP Mutations Identified by Whole-Exome Sequencing in a Norwegian Patient with Sporadic Ataxia and Edema. <i>Cerebellum</i> , 2017, 16, 272-275.	1.4	17
10	Characterization of DNA Substrate Binding to the Phosphatase Domain of the DNA Repair Enzyme Polynucleotide Kinase/Phosphatase. <i>Biochemistry</i> , 2017, 56, 1737-1745.	1.2	2
11	Systematic review of autosomal recessive ataxias and proposal for a classification. <i>Cerebellum and Ataxias</i> , 2017, 4, 3.	1.9	49
12	Ataxia with oculomotor apraxia is associated with the DNA damage repair pathway. <i>Movement Disorders</i> , 2017, 32, 720-720.	2.2	1
13	Coordination of DNA single strand break repair. <i>Free Radical Biology and Medicine</i> , 2017, 107, 228-244.	1.3	179
14	Novel <i>PNKP</i> mutation in siblings with ataxia-oculomotor apraxia type 4. <i>Journal of Neurogenetics</i> , 2017, 31, 23-25.	0.6	16
15	DNA repair in the trinucleotide repeat disorders. <i>Lancet Neurology</i> , The, 2017, 16, 88-96.	4.9	75
16	XRCC1 mutation is associated with PARP1 hyperactivation and cerebellar ataxia. <i>Nature</i> , 2017, 541, 87-91.	13.7	209
17	Genome integrity and disease prevention in the nervous system. <i>Genes and Development</i> , 2017, 31, 1180-1194.	2.7	117
18	Comparing ataxias with oculomotor apraxia: a multimodal study of AOA1, AOA2 and AT focusing on video-oculography and alpha-fetoprotein. <i>Scientific Reports</i> , 2017, 7, 15284.	1.6	21

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19	Neurological disorders associated with DNA strand-break processing enzymes. <i>Mechanisms of Ageing and Development</i> , 2017, 161, 130-140.	2.2	39
20	Polynucleotide kinase-phosphatase (PNKP) mutations and neurologic disease. <i>Mechanisms of Ageing and Development</i> , 2017, 161, 121-129.	2.2	48
21	The Rev1 interacting region (RIR) motif in the scaffold protein XRCC1 mediates a low-affinity interaction with polynucleotide kinase/phosphatase (PNKP) during DNA single-strand break repair. <i>Journal of Biological Chemistry</i> , 2017, 292, 16024-16031.	1.6	16
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27	DNA damage response. <i>Neurology</i> , 2018, 90, 367-376.	1.5	6
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29	Genetic Dystonia-ataxia Syndromes: Clinical Spectrum, Diagnostic Approach, and Treatment Options. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 373-382.	0.8	21
30	Two patients with <i>PNKP</i> mutations presenting with microcephaly, seizure, and oculomotor apraxia. <i>Clinical Genetics</i> , 2018, 93, 931-933.	1.0	10
31	Ataxia with oculomotor apraxia type 2: an evolving axonal neuropathy. <i>Practical Neurology</i> , 2018, 18, 52-56.	0.5	5
32	DNA repair in trinucleotide repeat ataxias. <i>FEBS Journal</i> , 2018, 285, 3669-3682.	2.2	12
33	The genetic nomenclature of recessive cerebellar ataxias. <i>Movement Disorders</i> , 2018, 33, 1056-1076.	2.2	61
34	Diseases Associated with Mutation of Replication and Repair Proteins. <i>Advances in Experimental Medicine and Biology</i> , 2018, 1076, 215-234.	0.8	5
35	Drosophila Models for Human Diseases. <i>Advances in Experimental Medicine and Biology</i> , 2018, , .	0.8	13
36	Impact of DNA repair and stability defects on cortical development. <i>Cellular and Molecular Life Sciences</i> , 2018, 75, 3963-3976.	2.4	13
37	The polynucleotide kinase- $\epsilon$ -phosphatase gene (PNKP) is involved in Charcot-Marie-Tooth disease (CMT2B2) previously related to MED25. <i>Neurogenetics</i> , 2018, 19, 215-225.	0.7	31

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40	XRCC1 protein; Form and function. DNA Repair, 2019, 81, 102664.	1.3	105
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61	Polyglutamine spinocerebellar ataxias: emerging therapeutic targets. <i>Expert Opinion on Therapeutic Targets</i> , 2020, 24, 1099-1119.	1.5	8
62	Pathological mutations in PNKP trigger defects in DNA single-strand break repair but not DNA double-strand break repair. <i>Nucleic Acids Research</i> , 2020, 48, 6672-6684.	6.5	37
63	A Novel Homozygous Variant in the Fork-Head-Associated Domain of Polynucleotide Kinase Phosphatase in a Patient Affected by Late-Onset Ataxia With Oculomotor Apraxia Type 4. <i>Frontiers in Neurology</i> , 2019, 10, 1331.	1.1	5
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65	A Novel c.968C>T homozygous Mutation in the Polynucleotide Kinase 3'â€²â€²â€²Phosphatase Gene Related to the Syndrome of Microcephaly, Seizures, and Developmental Delay. <i>Journal of Pediatric Genetics</i> , 2021, 10, 164-172.	0.3	0
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89	Prenatal phenotype of PNKP-related primary microcephaly associated with variants affecting both the FHA and phosphatase domain. <i>European Journal of Human Genetics</i> , 2022, 30, 101-110.	1.4	3
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91	Ocular Motor Apraxia. <i>Contemporary Clinical Neuroscience</i> , 2019, , 451-470.	0.3	1
92	Diagnosis and Management of Ataxia-Telangiectasia in Resource-Limited Settings. <i>Journal of International Child Neurology Association</i> , 2020, 1, .	0.0	1
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99	Clinical and Genetic Characterization of Brazilian Patients with Ataxia and Oculomotor Apraxia. <i>Movement Disorders</i> , 2022, , .	2.2	2
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