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| **Table e-1.** Manuscripts identified in systematic literature review with patient numbers and patient classification | | | |
|  | **Reference** | **# of pts.** | **Class** |
| 1. | Alkufri F, Harrower T, Rahman Y, et al. Molybdenum cofactor deficiency presenting with a Parkinsonism-dystonia syndrome. *Mov Disord*. 2013;28(3):399-400. doi:10.1002/mds.25276 | 1 | 2 |
| 2. | Appignani BA, Kaye EM, Wolpert SM. CT and MR appearance of the brain in two children with molybdenum cofactor deficiency. *Am J Neuroradiol*. 1996;17(2):317-320. | 2 | 1 |
| 3. | Arenas M, Fairbanks LD, Vijayakumar K, Carr L, Escuredo E, Marinaki AM. An unusual genetic variant in the MOCS1 gene leads to complete missplicing of an alternatively spliced exon in a patient with molybdenum cofactor deficiency. *J Inherit Metab Dis*. 2009;32(4):560-569. doi:10.1007/s10545-009-1151-7 **\*\*\* Duplicated as patient #8 in Vijayakumar et al. 2001 and patient (C) in Mills et al. 2012** | 1 | 2 |
| 4. | Arnold GL, Greene CL, Patrick Stout J, Goodman SI. Molybdenum cofactor deficiency. *J Pediatr*. 1993;123(4):595-598. doi:10.1016/S0022-3476(05)80961-8 | 1 | 1 |
| 5. | Arslanoglu S, Yalaz M, Gökşen D, et al. Molybdenum cofactor deficiency associated with Dandy-Walker complex. *Brain Dev*. 2001;23(8):815-818. http://www.ncbi.nlm.nih.gov/pubmed/11720800. | 1 | 1 |
| 6. | Bakker HD, Abeling NG, ten Houten R, et al. Molybdenum cofactor deficiency can mimic postanoxic encephalopathy. *J Inherit Metab Dis*. 1993;16(5):900-901. http://www.ncbi.nlm.nih.gov/pubmed/8295412. | 1 | 1 |
| 7. | Bamforth FJ, Johnson JL, Davidson AGF, Wong LTK, Lockitch G, Applegarth DA. Biochemical investigation of a child with molybdenum cofactor deficiency. *Clin Biochem*. 1990;23(6):537-542. doi:10.1016/0009-9120(90)80046-L | 1 | 2 |
| 8. | Barbot C, Martins E, Vilarinho L, Dorche C, Cardoso ML. A mild form of infantile isolated sulphite oxidase deficiency. *Neuropediatrics*. 1995;26(6):322-324. doi:10.1055/s-2007-979783 | 1 | 2 |
| 9. | Basheer SN, Waters PJ, Lam CW, et al. Isolated sulfite oxidase deficiency in the newborn: Lactic acidaemia and leukoencephalopathy. *Neuropediatrics*. 2007;38(1):38-41. doi:10.1055/s-2007-981484  **\*\*\* Duplicated as patient (G) in Mills et al. 2012** | 1 | 1 |
| 10. | Bayram E, Topcu Y, Karakaya P, et al. Molybdenum cofactor deficiency: Review of 12 cases (MoCD and review). *Eur J Paediatr Neurol*. 2013;17(1):1-6. doi:10.1016/j.ejpn.2012.10.003 | 1 | 1 |
| 11. | Bindu PS, Christopher R, Mahadevan A, Bharath RD. Clinical and imaging observations in isolated sulfite oxidase deficiency. *J Child Neurol*. 2011;26(8):1036-1040. doi:10.1177/0883073811401399 | 2 | 1 |
| 12. | Blau N, De Klerk JBC, Thöny B, et al. Tetrahydrobiopterin loading test in xanthine dehydrogenase and molybdenum cofactor deficiencies. *Biochem Mol Med*. 1996;58(2):199-203. doi:10.1006/bmme.1996.0049 | 2 | 1 |
| 13. | Boles RG, Ment LR, Meyn MS, Horwich AL, Kratz LE, Rinaldo P. Short-term response to dietary therapy in molybdenum cofactor deficiency. *AnnNeurol*. 1993;34(0364-5134):742-744. | 1 | 1 |
| 14. | Bonioli E, DiStefano A, Palmieri A, et al. Combined deficiency of xanthine oxidase and sulphite oxidase due to a deficiency of molybdenum cofactor. *J Inherit Metab Dis*. 1996;19(5):700-701. http://www.ncbi.nlm.nih.gov/pubmed/8892030. | 1 | 1 |
| 15. | Bosley TM, Alorainy IA, Oystreck DT, et al. Neurologic injury in isolated sulfite oxidase deficiency. *Can J Neurol Sci*. 2014;41(1):42-48. <http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=emed12&NEWS=N&AN=71468038>.  **\*\*\* Patient #4 duplicated in Seidahmed et al. 2005** | 5 | 1 |
| 16. | Brown GK, Scholem RD, Croll HB, Wraith JE, McGill JJ. Sulfite oxidase deficiency: clinical, neuroradiologic, and biochemical features in two new patients. *Neurology*. 1989;39(2 Pt 1):252-257. http://www.ncbi.nlm.nih.gov/pubmed/2915798. | 2 | 1 |
| 17. | Brucknerova I, Behulova D, Bzduch V, Mach M, Dubovicky M, Ujhazy E. Newborn with neonatal form of molybdenum cofactor deficiency - the first patient in the Slovak Republic. *Neuro Endocrinol Lett*. 2010;31 Suppl 2:5-7. http://www.ncbi.nlm.nih.gov/pubmed/21187823. | 1 | 1 |
| 18. | Carmi-Nawi N, Malinger G, Mandel H, Ichida K, Lerman-Sagie T, Lev D. Prenatal brain disruption in molybdenum cofactor deficiency. *J Child Neurol*. 2011;26(4):460-464. doi:10.1177/0883073810383017 | 1 | 1 |
| 19. | Carragher FM, Kirk JM, Steer C, Allen J, Dorche C. False negative thiosulphate screening test in a case of molybdenum cofactor deficiency. *J Inherit Metab Dis*. 1999;22(7):842-843. http://www.ncbi.nlm.nih.gov/pubmed/10518287. | 1 | 1 |
| 20. | Chan KY, Li CK, Lai CK, Ng SF, Chan AYW. Infantile isolated sulphite oxidase deficiency in a Chinese family: a rare neurodegenerative disorder. *Hong Kong Med J*. 2002;8(4):279-282. <http://www.ncbi.nlm.nih.gov/pubmed/12167732>. **\*\*\*Patient clinical data**  Lam C-W, Li C-K, Lai C-K, et al. DNA-based diagnosis of isolated sulfite oxidase deficiency by denaturing high-performance liquid chromatography. *Mol Genet Metab*. 2002;75(1):91-95. doi:10.1006/mgme.2001.3267 **\*\*\*Report of patient’s genotype** | 1 | 2 |
| 21. | Chen L-WW, Tsai Y-SS, Huang C-CC. Prenatal multicystic encephalopathy in isolated sulfite oxidase deficiency with a novel mutaion. *Pediatr Neurol*. 2014;51(1):181-182. doi:10.1016/j.pediatrneurol.2014.03.010 | 1 | 1 |
| 22. | Cho SY, Goh DLM, Lau KC, Ong HT, Lam C wan. Microarray analysis unmasked paternal uniparental disomy of chromosome 12 in a patient with isolated sulfite oxidase deficiency. *Clin Chim Acta*. 2013;426:13-17. doi:10.1016/j.cca.2013.08.013 | 1 | 1 |
| 23. | Coskun T, Yetuk M, Yurdakok M, Tekinalp G. Blood uric acid as a pointer to the diagnosis of molybdenum cofactor deficiency [4]. *Acta Paediatr Int J Paediatr*. 1998;87(6):714-715. doi:10.1080/080352598750014229 | 1 | 1 |
| 24. | Del Rizzo M, Burlina AP, Sass JO, et al. Metabolic stroke in a late-onset form of isolated sulfite oxidase deficiency. *Mol Genet Metab*. 2013;108(4):263-266. doi:10.1016/j.ymgme.2013.01.011 | 1 | 2 |
| 25. | Desjacques P, Mousson B, Vianey-Liaud C, et al. Combined deficiency of xanthine oxidase and sulfite oxidase: diagnosis of a new case followed by an antenatal diagnosis. *J Inher Metab Dis*. 1985;8 Suppl. 2:117-118. | 1 | 1 |
| 26. | Dublin AB, Hald JK, Wootton-Gorges SL. Isolated sulfite oxidase deficiency: MR imaging features. *Am J Neuroradiol*. 2002;23(3):484-485. | 1 | 1 |
| 27. | Duran M, Beemer FA, van de Heiden C, et al. Combined deficiency of xanthine oxidase and sulphite oxidase: a defect of molybdenum metabolism or transport? *J Inherit Metab Dis*. 1978;1(4):175-178. http://www.ncbi.nlm.nih.gov/pubmed/17254. | 1 | 1 |
| 28. | Edwards MC, Johnson JL, Marriage B, et al. Isolated sulfite oxidase deficiency: Review of two cases in one family. *Ophthalmology*. 1999;106(10):1957-1961. doi:10.1016/S0161-6420(99)90408-6 | 2 | 1 |
| 29. | Edwards M, Roeper J, Allgood C, et al. Investigation of molybdenum cofactor deficiency due to MOCS2 deficiency in a newborn baby. *Meta Gene*. 2015;3:43-49. doi:10.1016/j.mgene.2014.12.003 | 1 | 1 |
| 30. | Endres W, Shin YS, Günther R, Ibel H, Duran M, Wadman SK. Report on a new patient with combined deficiencies of sulphite oxidase and xanthine dehydrogenase due to molybdenum cofactor deficiency. *Eur J Pediatr*. 1988;148(3):246-249. http://www.ncbi.nlm.nih.gov/pubmed/3215199. | 1 | 1 |
| 31. | Eyaid WM, Al-Nouri DM, Rashed MS, Al-Rifai MT, Al-Wakeel AS. An inborn error of metabolism presenting as hypoxic-ischemic insult. *Pediatr Neurol*. 2005;32(2):134-136. doi:10.1016/j.pediatrneurol.2004.07.010 | 2 | 1 |
| 32. | Garrett RM, Johnson JL, Graf TN, Feigenbaum A, Rajagopalan K V. Human sulfite oxidase R160Q: Identification of the mutation in a sulfite oxidase-deficient patient and expression and characterization of the mutant enzyme. *Proc Natl Acad Sci*. 1998;95(11):6394-6398. doi:10.1073/pnas.95.11.6394 | 1 | 2 |
| 33. | Goh A, Lim KW. Sulphite oxidase deficiency--a report of two siblings. *Singapore Med J*. 1997;38(9):391-394. http://www.ncbi.nlm.nih.gov/pubmed/9407766. | 2 | 2 |
| 34. | Graf WD, Oleinik OE, Jack RM, Weiss AH, Johnson JL. Ahomocysteinemia in molybdenum cofactor deficiency. *Neurology*. 1998;51(3):860-862. http://www.ncbi.nlm.nih.gov/pubmed/9748040. | 1 | 2 |
| 35. | Gray RG, Green A, Basu SN, et al. Antenatal diagnosis of molybdenum cofactor deficiency. *Am J Obstet Gynecol*. 1990;163(4 Pt 1):1203-1204. http://www.ncbi.nlm.nih.gov/pubmed/2220930. | 1 | 1 |
| 36. | Gümüş H, Ghesquiere S, Per H, et al. Maternal uniparental isodisomy is responsible for serious molybdenum cofactor deficiency. *Dev Med Child Neurol*. 2010;52(9):868-872. doi:10.1111/j.1469-8749.2010.03724.x | 1 | 1 |
| 37. | Hansen LK, Wulff K, Dorche C, Christensen E. Molybdenum cofactor deficiency in two siblings: Diagnostic difficulties. *Eur J Pediatr*. 1993;152(8):662-664. doi:10.1007/BF01955243 | 2 | 1 |
| 38. | Higuchi R, Sugimoto T, Tamura A, et al. Early features in neuroimaging of two siblings with molybdenum cofactor deficiency. *Pediatrics*. 2014;133(1):e267-71. doi:10.1542/peds.2013-0935 | 2 | 1 |
| 39. | Hobson EE, Thomas S, Crofton PM, Murray AD, Dean JCS, Lloyd D. Isolated sulphite oxidase deficiency mimics the features of hypoxic ischaemic encephalopathy. *Eur J Pediatr*. 2005;164(11):655-659. doi:10.1007/s00431-005-1729-5 | 2 | 1 |
| 40. | Hoffmann C, Ben-Zeev B, Anikster Y, et al. Magnetic resonance imaging and magnetic resonance spectroscopy in isolated sulfite oxidase deficiency. *J Child Neurol*. 2007;22(10):1214-1221. doi:10.1177/0883073807306260 | 3 | 1 |
| 41. | Holder JL, Agadi S, Reese W, Rehder C, Quach MM. Infantile spasms and hyperekplexia associated with isolated sulfite oxidase deficiency. *JAMA Neurol*. 2014;71(6):782-784. doi:10.1001/jamaneurol.2013.5083 | 1 | 1 |
| 42. | Huang Y-L, Lin D-S, Huang J-K, Chiu N-C, Ho C-S. 99mTc-ethyl cysteinate dimer cranial single-photon emission computed tomography and serial cranial magnetic resonance imaging in a girl with isolated sulfite oxidase deficiency. *Pediatr Neurol*. 2012;47(1):44-46. doi:10.1016/j.pediatrneurol.2012.03.012 | 1 | 1 |
| 43. | Hughes EF, Fairbanks L, Simmonds HA, Robinson RO. Molybdenum cofactor deficiency - Phenotypic variability in a family with a late-onset variant. *Dev Med Child Neurol*. 1998;40(1):57-61. <http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=emed4&NEWS=N&AN=1998025251>.  **\*\*\*Patient #2 duplicated in Alkufri et al. 2013.** | 1 | 2 |
| 44. | Huijmans JGM, Schot R, de Klerk JBC, et al. Molybdenum cofactor deficiency: Identification of a patient with homozygote mutation in the MOCS3 gene. *Am J Med Genet Part A*. 2017;173(6):1601-1606. doi:10.1002/ajmg.a.38240 | 1 | 2 |
| 45. | Ichida K, Aydin HI, Hosoyamada M, et al. A Turkish case with molybdenum cofactor deficiency. *Nucleosides, Nucleotides and Nucleic Acids*. 2006;25(9-11):1087-1091. doi:10.1080/15257770600894022 | 1 | 2 |
| 46. | Irreverre F, Mudd SHH, Heizer WD, Laster L. Sulfite oxidase deficiency: Studies of a patient with mental retardation, dislocated ocular lenses, and abnormal urinary excretion of S-sulfo-l-cysteine, sulfite, and thiosulfate. *Biochem Med*. 1967;1(2):187-217. doi:10.1016/0006-2944(67)90007-5 | 1 | 1 |
| 47. | Johnson JL, Waud WR, Rajagopalan K V, Duran M, Beemer FA, Wadman SK. Inborn errors of molybdenum metabolism: combined deficiencies of sulfite oxidase and xanthine dehydrogenase in a patient lacking the molybdenum cofactor. *Proc Natl Acad Sci U S A*. 1980;77(6):3715-3719. doi:10.1073/pnas.77.6.3715 | 1 | 1 |
| 48. | Johnson JL, Coyne KE, Rajagopalan K V., et al. Molybdopterin synthase mutations in a mild case of molybdenum cofactor deficiency. *Am J Med Genet*. 2001;104(2):169-173. doi:10.1002/1096-8628(20011122)104:2<169::AID-AJMG1603>3.0.CO;2-8 | 1 | 2 |
| 49. | Johnson JL, Rajagopalan K V., Renier WO, Van Der Burgt I, Ruitenbeek W. Isolated sulfite oxidase deficiency: Mutation analysis and DNA-based prenatal diagnosis. *Prenat Diagn*. 2002;22(5):433-436. doi:10.1002/pd.335 | 1 | 1 |
| 50. | Kang PB, Kaye EM, Hunter J V., Kaye EM. Lactic acid elevation in extramitochondrial childhood neurodegenerative diseases. *J Child Neurol*. 2001;16(9):657-660. doi:10.1177/088307380101600906 | 1 | 1 |
| 51. | Kikuchi K, Hamano SI, Mochizuki H, Ichida K, Ida H. Molybdenum Cofactor Deficiency Mimics Cerebral Palsy: Differentiating Factors for Diagnosis. *Pediatr Neurol*. 2012;47(2):147-149. doi:10.1016/j.pediatrneurol.2012.04.013 | 1 | 1 |
| 52. | Lee HF, Chi CS, Tsai CR, Chen HC, Lee IC. Prenatal brain disruption in isolated sulfite oxidase deficiency. *Orphanet J Rare Dis*. 2017;12(1):1-5. doi:10.1186/s13023-017-0668-3 | 1 | 1 |
| 53. | Macaya A, Brunso L, Fernández-Castillo N, et al. Molybdenum cofactor deficiency presenting as neonatal hyperekplexia: A clinical, biochemical and genetic study. *Neuropediatrics*. 2005;36(6):389-394. doi:10.1055/s-2005-872877 | 1 | 1 |
| 54. | Megahed H, Nicouleau M, Barcia G., et al. Utility of whole exome sequencing for the early diagnosis of pediatric-onset cerebellar atrophy associated with developmental delay in an inbred population. *Orphanet Journal of Rare Diseases.* 2016;11:57. Doi:10.1186/s13023-016-0436-9. | 1 | 2 |
| 55. | Mills PB, Footitt EJ, Ceyhan S, et al. Urinary AASA excretion is elevated in patients with molybdenum cofactor deficiency and isolated sulphite oxidase deficiency. *J Inherit Metab Dis*. 2012;35(6):1031-1036. doi:10.1007/s10545-012-9466-1 **\*\*\* Patient (C) duplicated in patient #8 in Vijayakumar et al. and Arenas et al. 2009** | 6/1 | 1/2 |
| 56. | Mize C, Johnson JL, Rajagopalan K V. Defective molybdopterin biosynthesis: clinical heterogeneity associated with molybdenum cofactor deficiency. *J Inherit Metab Dis*. 1995;18(3):283-290. doi:10.1007/BF00710416 | 1 | 2 |
| 57. | Nagappa M, Bindu PS, Taly AB, Sinha S, Bharath RD. Child Neurology: Molybdenum cofactor deficiency. *Neurology*. 2015;85(23):e175-e178. doi:10.1212/WNL.0000000000002194 | 1 | 1 |
| 58. | Ngu LH, Afroze B, Chen BC, Affandi O, Zabedah MY. Molybdenum cofactor deficiency in a Malaysian child. *Singapore Med J*. 2009;50(10):365-367. | 1 | 1 |
| 59. | Parini R, Briscioli V, Caruso U, et al. Spherophakia associated with molybdenum cofactor deficiency. *Am J Med Genet*. 1997;73(3):272-275. http://www.ncbi.nlm.nih.gov/pubmed/9415683. | 1 | 1 |
| 60. | Per H, Gümüş H, Ichida K, Çaǧlayan O, Kumandaş S. Molybdenum cofactor deficiency: Clinical features in a Turkish patient. *Brain Dev*. 2007;29(6):365-368. doi:10.1016/j.braindev.2006.10.007 | 1 | 1 |
| 61. | Pintos-Morell G, Naranjo MA, Artigas M, et al. Molybdenum cofactor deficiency associated with Dandy-Walker malformation. *J Inherit Metab Dis*. 1995;18(1):86-87. http://www.ncbi.nlm.nih.gov/pubmed/7623453. | 1 | 1 |
| 62. | Reiss J, Christensen E, Dorche C. Molybdenum cofactor deficiency: first prenatal genetic analysis. *Prenat Diagn*. 1999;19(4):386-388. http://www.ncbi.nlm.nih.gov/pubmed/10327149. | 2 | 1 |
| 63. | Reiss J, Gross-Hardt S, Christensen E, Schmidt P, Mendel RR, Schwarz G. A mutation in the gene for the neurotransmitter receptor-clustering protein gephyrin causes a novel form of molybdenum cofactor deficiency. *Am J Hum Genet*. 2001;68(1):208-213. doi:10.1086/316941 | 1 | 1 |
| 64. | Reiss J, Lenz U, Aquaviva-Bourdain C, Joriot-Chekaf S, Mention-Mulliez K, Holder-Espinasse M. A GPHN point mutation leading to molybdenum cofactor deficiency. *Clin Genet*. 2011;80(6):598-599. doi:10.1111/j.1399-0004.2011.01709.x | 1 | 1 |
| 65. | Rocha S, Ferreira AC, Dias AI, Vieira JP, Sequeira S. Sulfite oxidase deficiency - An unusual late and mild presentation. *Brain Dev*. 2014;36(2):176-179. doi:10.1016/j.braindev.2013.01.013 | 1 | 2 |
| 66. | Rupar CA, Gillett J, Gordon BA, et al. Isolated sulfite oxidase deficiency. *Neuropediatrics*. 1996;27(6):299-304. doi:10.1055/s-2007-973798 | 1 | 1 |
| 67. | Salman MS, Ackerley C, Senger C, Becker L. New insights into the neuropathogenesis of molybdenum cofactor deficiency. *Can J Neurol Sci*. 2002;29(1):91-96. doi:10.1017/S0317167100001803 | 1 | 1 |
| 68. | Salvan AM, Chabrol B, Lamoureux S, Confort-Gouny S, Cozzone PJ, Vion-Dury J. In vivo brain proton MR spectroscopy in a case of molybdenum cofactor deficiency. *Pediatr Radiol*. 1999;29(11):846-848. doi:10.1007/s002470050710 | 1 | 1 |
| 69. | Sass JO, Gunduz A, Araujo Rodrigues Funayama C, et al. Functional deficiencies of sulfite oxidase: Differential diagnoses in neonates presenting with intractable seizures and cystic encephalomalacia. *Brain Dev*. 2010;32(7):544-549. doi:10.1016/j.braindev.2009.09.005 | 2 | 1 |
| 70. | Schiaffino MC, Fantasia AR, Minniti G, Caruso U, Carnevale F, Cerone R. Isolated sulphite oxidase deficiency: clinical and biochemical features in an Italian patient. *J Inherit Metab Dis*. 2004;27(1):101-102. doi:10.1023/B:BOLI.0000016674.61073.e4 | 1 | 1 |
| 71. | Schuierer G, Kurlemann G, Bick U, Stephani U. Molybdenum-cofactor deficiency: CT and MR findings. *Neuropediatrics*. 1995;26(1):51-54. doi:10.1055/s-2007-979720 | 4 | 1 |
| 72. | Schwahn BC, Van Spronsen FJ, Belaidi AA, et al. Efficacy and safety of cyclic pyranopterin monophosphate substitution in severe molybdenum cofactor deficiency type A: A prospective cohort study. *Lancet*. 2015;386(10007):1955-1963. doi:10.1016/S0140-6736(15)00124-5 **\*\*\* All MoCD type A patients treated with cPMP were excluded.** | 7 | 1 |
| 73. | Seidahmed MZ, Alyamani EA, Rashed MS, et al. Total truncation of the molybdopterin/dimerization domains of SUOX protein in an Arab family with isolated sulfite oxidase deficiency. *Am J Med Genet*. 2005;136 A(2):205-209. doi:10.1002/ajmg.a.30796 **\*\*\* Duplicated in Bosley et al. 2014** | 1 | 1 |
| 74. | Serrano M, Lizarraga I, Reiss J, et al. Cranial ultrasound and chronological changes in molybdenum cofactor deficiency. *Pediatr Radiol*. 2007;37(10):1043-1046. doi:10.1007/s00247-007-0558-2 | 1 | 1 |
| 75. | Shih VE, Abroms IF, Johnson JL, et al. Sulfite oxidase deficiency. Biochemical and clinical investigations of a hereditary metabolic disorder in sulfur metabolism. *N Engl J Med*. 1977;297(19):1022-1028. doi:10.1056/NEJM197711102971902 | 1 | 2 |
| 76. | Sie SD, De Jonge RCJ, Blom HJ, et al. Chronological changes of the amplitude-integrated EEG in a neonate with molybdenum cofactor deficiency. *J Inherit Metab Dis*. 2010;33(SUPPL. 3):401-407. doi:10.1007/s10545-010-9198-z | 1 | 1 |
| 77. | Slot HMJ, Overweg-Plandsoen WCG, Bakker HD, et al. Molybdenum-cofactor deficiency: An easily missed cause of neonatal convulsions. *Neuropediatrics*. 1993;24(3):139-142. doi:10.1055/s-2008-1071531 | 2 | 1 |
| 78. | Stence N V., Coughlin CR, Fenton LZ, Thomas JA. Distinctive pattern of restricted diffusion in a neonate with molybdenum cofactor deficiency. *Pediatr Radiol*. 2013;43(7):882-885. doi:10.1007/s00247-012-2579-8 | 1 | 1 |
| 79. | Struys EA, Nota B, Bakkali A, Al Shahwan S, Salomons GS, Tabarki B. Pyridoxine-dependent Epilepsy With Elevated Urinary  -Amino Adipic Semialdehyde in Molybdenum Cofactor Deficiency. *Pediatrics*. 2012;130(6):e1716-e1719. doi:10.1542/peds.2012-1094 | 2 | 1 |
| 80. | Tan W-HW-H, Eichler FS, Hoda S, et al. Isolated sulfite oxidase deficiency: a case report with a novel mutation and review of the literature. *Pediatrics*. 2005;116(3):757-766. doi:10.1542/peds.2004-1897 | 1 | 1 |
| 81. | Tardy P, Parvy P, Charpentier C, Bonnefont J, Saudubray J. CASE REPORT Attempt at therapy in sulphite oxidase deficiency Sulphite oxidase deficiency ( McKusick 27230 ) is an hereditary disorder of sulphur metabolism . The degradative metabolism of cysteine and methionine in normal individuals entails the conversi. 1989;12:94-95. | 1 | 2 |
| 82. | Teksam O, Yurdakok M, Coskun T. Molybdenum cofactor deficiency presenting with severe metabolic acidosis and intracranial hemorrhage. *J Child Neurol*. 2005;20(2):155-157. http://www.ncbi.nlm.nih.gov/pubmed/15794186. | 1 | 1 |
| 83. | Tezel G, Oztekin O, Kalay S, Aslan A, Akçakuş M, Oygür N. The association of molybdenum cofactor deficiency and pyloric stenosis. *J Perinatol*. 2012;32(11):896-898. doi:10.1038/jp.2011.192 | 1 | 1 |
| 84. | Topcu M, Coskun T, Haliloglu G, Saatci I. Molybdenum cofactor deficiency: Report of three cases presenting as hypoxic-ischemic encephalopathy. *J Child Neurol*. 2001;16(4):264-270. doi:10.1177/088307380101600406 | 3 | 1 |
| 85. | Touati G, Rusthoven E, Depondt E, et al. Dietary therapy in two patients with a mild form of sulphite oxidase deficiency. Evidence for clinical and biological improvement. *J Inherit Metab Dis*. 2000;23(1):45-53. http://www.ncbi.nlm.nih.gov/pubmed/10682307. | 2 | 2 |
| 86. | van der Klei-van Moorsel JM, Smit LME, Brockstedt M, Jakobs C, Dorche C, Duran M. Infantile isolated sulphite oxidase deficiency: Report of a case with negative sulphite test and normal sulphate excretion. *Eur J Pediatr*. 1991;150(3):196-197. doi:10.1007/BF01963565 | 1 | 2 |
| 87. | van Gennip AH, Abeling NG, Stroomer SAE, Overmarks H, Bakker HD. The detection of molybdenum cofactor deficiency: clinical symptomatology and urinary metabolite profile. *J Inherit Metab Dis*. 1994;17(1964):142-145. | 2 | 1 |
| 88. | Veldman A, Santamaria-Araujo JA, Sollazzo S, et al. Successful Treatment of Molybdenum Cofactor Deficiency Type A With cPMP. *Pediatrics*. 2010;125(5):e1249-e1254. doi:10.1542/peds.2009-2192 | 1 | 1 |
| 89. | Vianey-Liaud C, Desjacques P, Gaulme J, et al. A new case of isolated sulphite oxidase deficiency with rapid fatal outcome. *J Inherit Metab Dis*. 1988;11(4):425-426. http://www.ncbi.nlm.nih.gov/pubmed/3149702. | 1 | 1 |
| 90. | Vijayakumar K, Gunny R, Grunewald S, et al. Clinical neuroimaging features and outcome in molybdenum cofactor deficiency. *Pediatr Neurol*. 2011;45(4):246-252. doi:10.1016/j.pediatrneurol.2011.06.006 **\*\*\* Patinet #8 duplicates in Arenas et al. 2009 and as patient (C) in Mills et al. 2012** | 6/1 | 1/2 |
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