

INBORN ERRORS OF METABOLISM - I

NEUROIMAGING IN INBORN ERRORS OF METABOLISM - DIAGNOSTIC PATTERN AND CLINICAL IMPLICATIONS

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Abstract: *Inborn errors of metabolism often produce characteristic, symmetric brain injury patterns that can be recognized on Magnetic Resonance Imaging and Magnetic Resonance Spectroscopy. A systematic, pattern-based approach-integrating patient age, clinical evolution, and tissue predilection-enables early and accurate diagnosis. Distinct imaging signatures such as elevated N-acetylaspartate in Canavan disease or restricted diffusion in Maple syrup urine disease directly warrant urgent management and prognostication. Advances in neuroimaging now guide gene and transplant-based therapies, underscoring its pivotal role in the early recognition and treatment of paediatric neurometabolic disorders.*

Keywords: *Pediatric neuroimaging, Inborn errors of metabolism, Magnetic resonance spectroscopy, Pattern recognition.*

Points to Remember

- *Selective vulnerability of brain structures helps to record specific images which point towards specific metabolic disorders.*
- *Typical MRI patterns can predict certain specific white matter disorders.*
- *Patterns do not always confirm to specific disorders, since at early stages / subclinical and burnt-out stages of the disorders, patterns are not typical on MRI.*
- *MR Spectroscopy is useful for the confirmation of some of the metabolic disorders.*

Bibliography

1. Liserre R, Pinelli L, Gasparotti R. MR spectroscopy in pediatric neuroradiology. *Transl Pediatr.* 2021 Apr;10(4):1169-1200. doi: 10.21037/tp-20-445. PMID: 34012861; PMCID: PMC8107850. <https://pubmed.ncbi.nlm.nih.gov/34012861/>
2. Gaillard F, Knipe H, Buemi F, et al. Canavan disease. *Radiopaedia.org.* Accessed 24 Oct 2025. <https://doi.org/10.53347/rID-1045>
3. Barkovich AJ, Raybaud C. *Pediatric neuroimaging.* 6th ed. New York: Thieme; 2019. <https://hsrc.himmelfarb.gwu.edu/books/121>
4. Wolf NI, Engelen M, van der Knaap MS. MRI pattern recognition in white matter disease. *Handb Clin Neurol.* 2024;204:37-50. doi: 10.1016/B978-0-323-99209-1.00019-3. PMID: 39322391. <https://pubmed.ncbi.nlm.nih.gov/39322391/>
5. Aida N. 1H-MR spectroscopy of the early developmental brain, neonatal encephalopathies, and neurometabolic disorders. *Magn Reson Med Sci.* 2022 Mar 1; 21(1): 9-28. doi: 10.2463/mrms.rev.2021-0055. Epub 2021 Aug 21. PMID: 34421092; PMCID: PMC9199977. <https://pubmed.ncbi.nlm.nih.gov/34421092/>

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6. Cornelius LP, Kannan B, Saravanan V, Venkatesan EP. Intramyelinic edema in maple syrup urine disease. *Ann Indian Acad Neurol.* 2014 Apr; 17(2):211-3. doi: 10.4103/0972-2327.132638. PMID: 25024577; PMCID: PMC4090852. <https://pubmed.ncbi.nlm.nih.gov/25024577/>
7. La Piana R, Uggetti C, Roncarolo F, Vanderver A, Olivieri I, Tonduti D, et al. Neuroradiologic patterns and novel imaging findings in Aicardi-Goutières syndrome. *Neurology.* 2016 Jan 5; 86(1):28-35. doi: 10.1212/WNL.0000000000002228. Epub 2015 Nov 18. PMID: 26581299; PMCID: PMC4731289. <https://pubmed.ncbi.nlm.nih.gov/26581299/>
8. Schiffmann R, van der Knaap MS. Invited article: an MRI-based approach to the diagnosis of white matter disorders. *Neurology.* 2009 Feb 24; 72(8):750-9. doi: 10.1212/01.wnl.0000343049.00540.c8. PMID: 19237705; PMCID: PMC2677542. <https://pubmed.ncbi.nlm.nih.gov/19237705/>
9. Jha P, Elfeky M, Fortin F, et al. Leopard skin sign (white matter). *Radiopaedia.org.* Accessed 24 Oct 2025. <https://doi.org/10.53347/rID-17588>
10. Reis E, Coolen T, Lolli V. MRI findings in acute hyperammonemic encephalopathy: three cases of different etiologies. *J Belg Soc Radiol.* 2020 Jan 30;104(1):9. doi: 10.5334/jbsr.2017. PMID: 32025625; PMCID: PMC6993592. <https://pubmed.ncbi.nlm.nih.gov/32025625/>
11. Sehgal R, Sharma S, Sankhyan N, Kumar A, Gulati S. Selective corticospinal tract involvement in late-onset Krabbe disease. *Neurology.* 2011 Jul 19;77(3):e20. doi: 10.1212/WNL.0b013e318225aaf5. PMID: 21768595. <https://pubmed.ncbi.nlm.nih.gov/21768595/>
12. Twomey EL, Naughten ER, Donoghue VB, Ryan S. Neuroimaging findings in glutaric aciduria type 1. *Pediatr Radiol.* 2003 Dec;33(12):823-30. doi: 10.1007/s00247-003-0956-z. Epub 2003 Oct 8. PMID: 14534757. <https://pubmed.ncbi.nlm.nih.gov/14534757/>
13. Loes DJ, Hite S, Moser H, Stillman AE, Shapiro E, Lockman L, et al. Adrenoleukodystrophy: a scoring method for brain MR observations. *AJNR Am J Neuroradiol.* 1994 Oct;15(9):1761-6. PMID: 7847225; PMCID: PMC8333737. <https://pubmed.ncbi.nlm.nih.gov/7847225/>
14. Bonfante E, Koenig MK, Adejumo RB, Perinjelil V, Riascos RF. The neuroimaging of Leigh syndrome: case series and review of the literature. *Pediatr Radiol.* 2016 Apr;46(4):443-51. doi: 10.1007/s00247-015-3523-5. Epub 2016 Jan 6. PMID: 26739140. <https://pubmed.ncbi.nlm.nih.gov/26739140/>
15. Kilicarslan R, Alkan A, Demirkol D, Toprak H, Sharifov R. Maple syrup urine disease: diffusion-weighted MRI findings during acute metabolic encephalopathic crisis. *Jpn J Radiol.* 2012 Jul; 30(6):522-5. doi: 10.1007/s11604-012-0079-2. Epub 2012 Apr 3. PMID: 22476847. <https://pubmed.ncbi.nlm.nih.gov/22476847/>
16. Kim JH, Lim MK, Jeon TY, Rha JH, Eo H, Yoo SY, et al. Diffusion and perfusion characteristics of MELAS (mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episode) in thirteen patients. *Korean J Radiol.* 2011 Jan-Feb;12(1):15-24. doi: 10.3348/kjr.2011.12.1.15. Epub 2011 Jan 3. Erratum in: *Korean J Radiol.* 2011 Mar;12(2):268. PMID: 21228936; PMCID: PMC3017880. <https://pubmed.ncbi.nlm.nih.gov/21228936/>
17. Takanashi J, Barkovich AJ, et al. Nonketotic hyperglycinemia: diffusion-weighted MRI pattern and glycine peak at 3.55 ppm on MR spectroscopy. *AJNR Am J Neuroradiol.* 2001 May; 22(5):872-7. doi: 10.3174/ajnr.A1383. PMID: 11309371. <https://pubmed.ncbi.nlm.nih.gov/11309371/>