

ON-LINE APPENDIX: REFERENCES OF LITERATURE

REVIEW

1. Agert F, Crippa AC, Lorenzoni PJ, et al. **Menkes' disease: case report.** *Arq Neuropsiquiatr* 2007;65:157–60 CrossRef Medline
2. Barnerias C, Boddaert N, Guiraud P, et al. **Unusual magnetic resonance imaging features in Menkes disease.** *Brain Dev* 2008;30:489–92 CrossRef Medline
3. Bekiesińska-Figatowska M, Rokicki D, Walecki J, et al. **Menkes' disease with a Dandy-Walker variant: case report.** *Neuroradiology* 2001;43:948–50 CrossRef Medline
4. Bernhard MK, Merkenschlager A, Mayer T, et al. **The spectrum of neuroradiological features in Menkes disease: widening of the cerebral venous sinuses.** *J Pediatr Neuroradiol* 2012;1:121–25
5. Bindu PS, Taly AB, Kothari S, et al. **Electro-clinical features and magnetic resonance imaging correlates in Menkes disease.** *Brain Dev* 2013;35:398–405 CrossRef Medline
6. Burgemeister AL, Zirn B, Oeffner F, et al. **Menkes disease with discordant phenotype in female monozygotic twins.** *Am J Med Genet A* 2015;167A:2826–29 CrossRef Medline
7. Choudhary R, Choudhary A, Sitaraman S. **Menkes disease—a rare neurodegenerative disorder.** *J Nepal Paediatr Soc* 2016;35:177–80 CrossRef
8. Cosimo QC, Daniela L, Elsa B, et al. **Kinky hair, kinky vessels, and bladder diverticula in Menkes disease.** *J Neuroimaging* 2011;21:e114–16 CrossRef Medline
9. Ekici B, Çaliskan M, Tatlı B. **Reversible temporal lobe edema: An early MRI finding in Menkes disease.** *J Pediatr Neurosci* 2012;7:160–61 CrossRef Medline
10. Faerber EN, Grover WD, DeFilipp GJ, et al. **Cerebral MR of Menkes kinky-hair disease.** *AJNR Am J Neuroradiol* 1989;10:190–92 Medline
11. Gandhi R, Kakkar R, Rajan S, et al. **Menkes kinky hair syndrome: a rare neurodegenerative disease.** *Case Rep Radiol* 2012;2012:684309 CrossRef Medline
12. Geller TJ, Pan Y, Martin DS. **Early neuroradiologic evidence of degeneration in Menkes' disease.** *Pediatr Neurol* 1997;17:255–58 CrossRef Medline
13. George S, Matthai SA, Sosamma MM, et al. **Menkes' kinky hair syndrome.** *Indian J Pediatr* 2005;72:891–92 CrossRef Medline
14. Hsich GE, Robertson RL, Irons M, et al. **Cerebral infarction in Menkes' disease.** *Pediatr Neurol* 2000;23:425–28 CrossRef Medline
15. Ito H, Mori K, Sakata M, et al. **Pathophysiology of the transient temporal lobe lesion in a patient with Menkes disease.** *Pediatr Int* 2008;50:825–27 CrossRef Medline
16. Jacobs DS, Smith AS, Finelli DA, et al. **Menkes kinky hair disease: characteristic MR angiographic findings.** *AJNR Am J Neuroradiol* 1993;14:1160–63 Medline
17. Jain P, Sharma S, Sankhyan N, et al. **Macrocephaly with diffuse white matter changes simulating a leukodystrophy in Menkes disease.** *Indian J Pediatr* 2013;80:160–62 CrossRef Medline
18. Jayawant S, Halpin S, Wallace S. **Menkes kinky hair disease: an unusual case.** *Eur J Paediatr Neurol* 2000;4:131–34 CrossRef Medline
19. Johnsen DE, Coleman L, Poe L. **MR of progressive neurodegenerative change in treated Menkes' kinky hair disease.** *Neuroradiology* 1991;33:181–82 CrossRef Medline
20. Datta KA, Ghosh T, Nayak K, et al. **Menkes kinky hair disease: a case report.** *Cases J* 2008;1:158 CrossRef Medline
21. Kim JH, Lee BH, Kim YM, et al. **Novel mutations and clinical outcomes of copper-histidine therapy in Menkes disease patients.** *Metab Brain Dis* 2015;30:75–81 CrossRef Medline
22. Kim OH, Suh JH. **Intracranial and extracranial MR angiography in Menkes disease.** *Pediatr Radiol* 1997;27:782–84 CrossRef Medline
23. Kim YH, Lee R, Yoo HW, et al. **Identification of a novel mutation in the ATP7A gene in a Korean patient with Menkes disease.** *J Korean Med Sci* 2011;26:951–53 CrossRef Medline
24. Koprivsek K, Lucic M, Kozic D, et al. **Basal ganglia lesions in the early stage of Menkes disease.** *J Inherit Metabol Dis* 2010;33:301–02 CrossRef
25. Lee ES, Ryoo JW, Choi DS, et al. **Diffusion-weighted MR imaging of unusual white matter lesion in a patient with Menkes disease.** *Korean J Radiol* 2007;8:82–85 CrossRef Medline
26. Leventer RJ, Kornberg AJ, Phelan EM, et al. **Early magnetic resonance findings in Menkes' disease.** *J Child Neurol* 1997;12:222–24 CrossRef Medline
27. Lin YJ, Ho CS, Hsu CH, et al. **A truncating de novo point mutation in a young infant with severe Menkes disease.** *Pediatr Neonatol* 2017;58:89–92 CrossRef Medline
28. Lubbe E. **From the coalface of clinical paediatric neurology: Menkes disease—a lesson not to be forgotten.** *S Afr J Child Health* 2012;6:56–59
29. Okada T, Sasaki F, Honda S, et al. **Menkes disease with gastroesophageal reflux disease and successful surgical treatment: a case report and literature.** *Turk J Pediatr* 2010;52:333–35 Medline
30. Ozawa H, Kodama H, Murata Y, et al. **Transient temporal lobe changes and a novel mutation in a patient with Menkes disease.** *Pediatr Int* 2001;43:437–40 CrossRef Medline
31. Fister P, Raku J, Primec RZ, et al. **Menkes kinky hair disease (Menkes syndrome): a case report.** *Acta Dermatovenerol Alp Pannonica Adriat* 2006;15:126–30 Medline
32. Park HD, Moon HK, Lee J, et al. **A novel ATP7A gross deletion mutation in a Korean patient with Menkes disease.** *Ann Clin Lab Sci* 2009;39:188–91 Medline
33. Pinto F, Calderazzi A, Canapicchi R, et al. **Radiological findings in a case of Menkes' disease.** *Child Nervous Syst* 1995;11:112–14 CrossRef Medline
34. Rego, JI, Rocha AJD, Segatelli V, et al. **Imaging features that allow for the recognition of Menkes disease.** *Arq Neuropsiquiatr* 2014;72:396 CrossRef Medline
35. Rennert J, Doelken R, Doelken M, et al. **Menkes disease: MRI appearance of a rare neurodegenerative disorder.** *J Pediatr Neurol* 2009;7:317–20
36. Rizk T, Mahmoud A, Jamali T, et al. **Menkes disease presenting with epilepsia partialis continua.** *Case Rep Neurol Med* 2014;2014:525784 CrossRef Medline
37. Saha S, Mridha D. **An unusual cause for focal convulsions: Menkes kinky hair disease.** *J Pediatr Neurol* 2013;11:123–25
38. Santos LM, Teixeira CD, Vilanova, LC, et al. **Menkes disease: case report of an uncommon presentation with white matter lesions.** *Arq Neuropsiquiatr* 2001;59:125–27 CrossRef Medline
39. Sener RN. **Menkes' disease (trichopoliodystrophy).** March 23, 2001. <https://www.eurorad.org/eurorad/case.php?id=968&teaching=true>. Accessed April 4, 2017 CrossRef
40. Seshadri R, Bindu PS, Gupta AK. **Teaching neuroimages: Menkes kinky hair syndrome.** *Neurology* 2013;81:12–13 CrossRef Medline
41. Sirleto P, Surace C, Santos H, et al. **Lyonization effects of the t(X;16) translocation on the phenotypic expression in a rare female with Menkes disease.** *Pediatr Res* 2009;65:347–51 CrossRef Medline
42. Smpokou P, Samanta M, Berry GT, et al. **Menkes disease in affected females: the clinical disease spectrum.** *Am J Med Genet A* 2015;167A:417–20 CrossRef Medline
43. Takahashi S, Ishii K, Matsumoto, K, et al. **Cranial MRI and MR angiography in Menkes' syndrome.** *Neuroradiology* 1993;35:556–58 CrossRef Medline
44. Thomas B, Dossary N, Widjaja E. **MRI of childhood epilepsy due to inborn errors of metabolism.** *AJR Am J Roentgenol* 2010;194:W367–74 CrossRef Medline
45. Venta-Sobero JA, Porras-Kattz E, Gutiérrez-Moctezuma J. **West syndrome as an epileptic presentation in Menkes' disease: two cases report [in Spanish].** *Rev Neurol* 2004;39:133–36 Medline
46. Menkes disease. Radiopaedia.org. <http://radiopaedia.org/cases/menkes-disease-1>. Accessed April 4, 2017
47. Zikou AK, Mouka V, Mpatsoulis A, et al. **Menkes disease: alteration of MR findings over time.** May 14, 2015. <https://www.eurorad.org/eurorad/case.php?id=12713&lang=en>. Accessed April 4, 2017 CrossRef

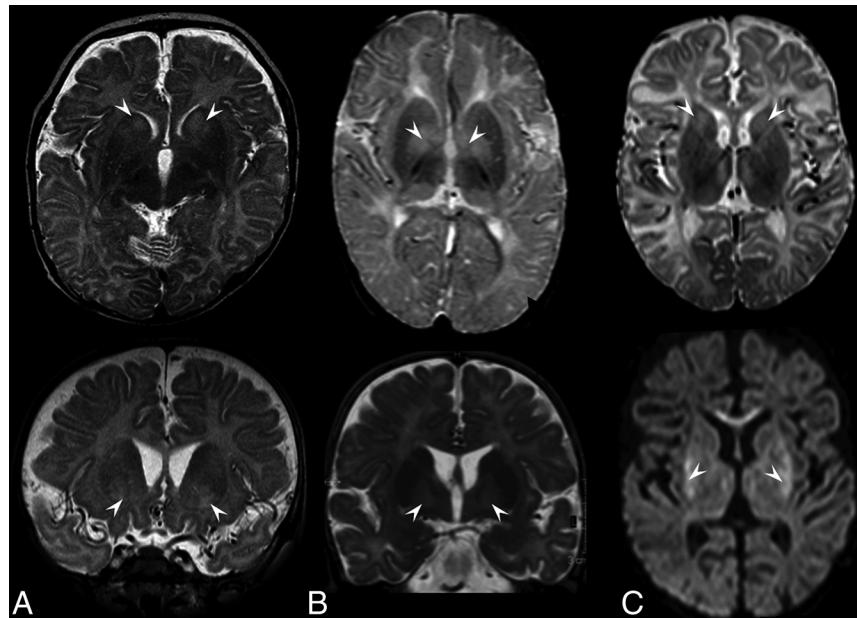
On-line Table: Demographics and main clinical data of our children with MD and children in the literature with MD who underwent brain MRI

Demographics	Present Sample	Literature Review (On-line Appendix)
Sample size	26 Children	62 Children
Sex	25 M; 1 F	57 M; 5 F
Age at clinical onset (mean) (range) (mo)	3.7 ± 2.5 (1–8) ^a	4.5 ± 3.3 (0.1–14) ^b
Age at first MRI (mean) (range) (mo)	7.5 ± 5.9 (0.3–32.2)	7.1 ± 5.6 (0.6–34.0)
Follow-up MRI	8/26 Children	23/62 Children
Age at last follow-up (mean) (range) (mo)	25.3 ± 19.4 (3.6–86.7)	10.2 ± 6.28 (1.5–30)
Follow-up duration (mean) (range) (mo)	22.8 ± 23.1 (1.4–78.5)	6.1 ± 5.9 (0.3–26)
Epilepsy	23/26	37/62
Age at epilepsy onset (mean) (range) (mo)	6.5 ± 3.4 (2.2–15.0)	4.6 ± 3.3 (0.1–14) ^c

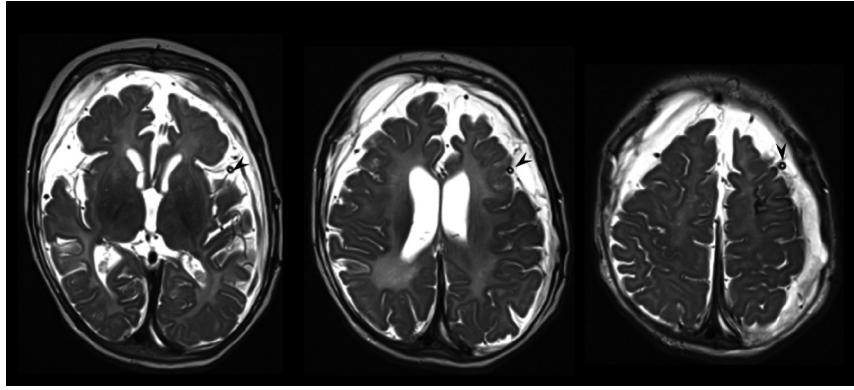
^a Calculated from 25/26 MD children.

^b Calculated from 31/62 MD children.

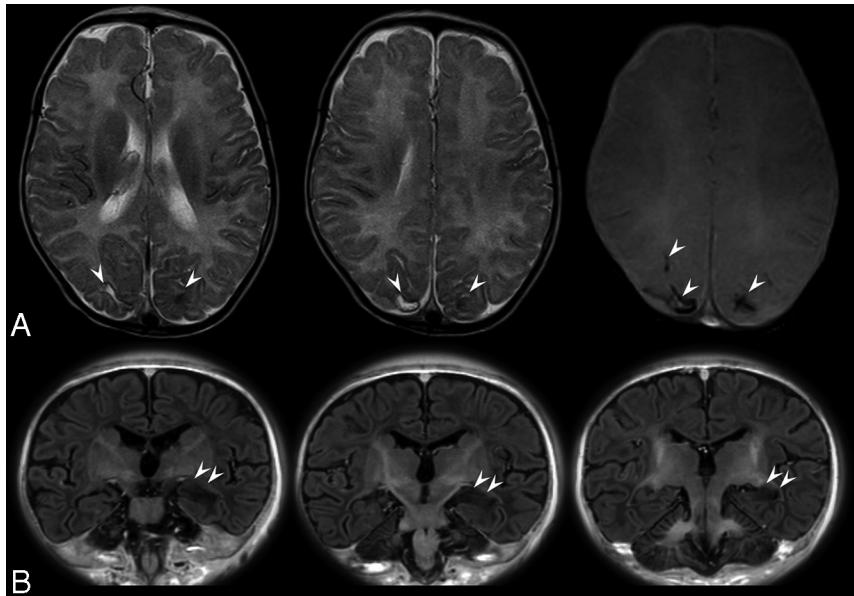
^c Calculated from 19/37 MD children with epilepsy.



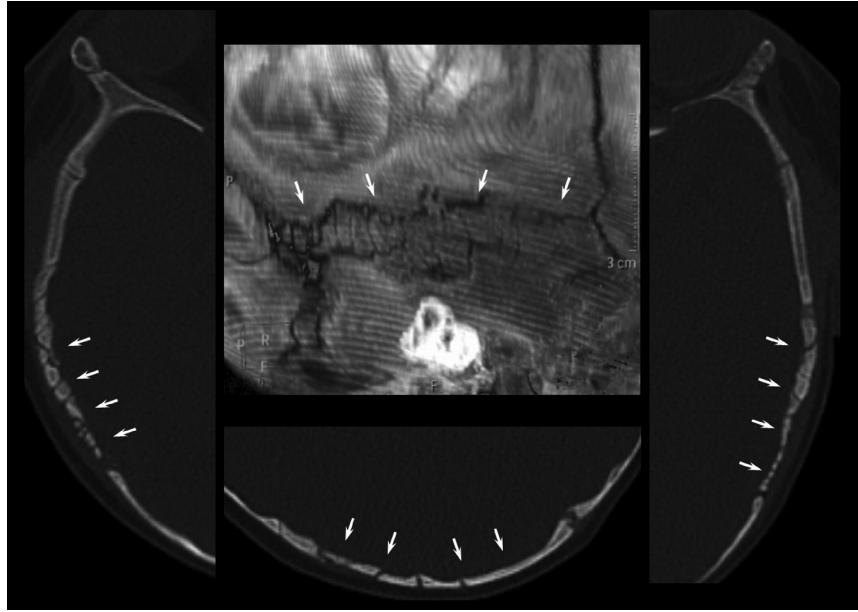
ON-LINE FIG 1. A, Axial and coronal T2-weighted images at the level of the basal ganglia showing asymmetric and inhomogeneous hyperintensity of the anterior neostriatum (arrowheads). Note the tortuosity of intracranial arteries and the mild supratentorial atrophy in a 5-month-old boy (patient 10) (normocephaly). B, Axial and coronal T2-weighted images at the level of the basal ganglia show bilateral symmetric hyperintensity of the globus pallidus (arrowheads) in a 4-month-old boy (patient 26). C, Axial T2- and diffusion-weighted images in a 14-month-old boy (patient 15) at the level of the basal ganglia show signal abnormality of the caudate head and medial thalamus bilaterally; the putamina appear slightly hyperintense on DWI (arrowheads). Note the hyperintensity of the genu of the corpus callosum in possible relationship with a recent status epilepticus.



ON-LINE FIG 2. Axial T2-weighted images in a 14-month-old boy (patient 14) showing bilateral subdural collections. Signal inhomogeneity and septa reveal different timing of the collections. Note brain atrophy (the child had microcephalia) and abnormal myelination that most likely are the combined result of delayed myelination and neurodegeneration. Cortical hemosiderin at the left superior frontal sulcus was due to a small iatrogenic blood clot (not shown) close to the intracranial end of a drainage (arrowheads).



ON-LINE FIG 3. *A*, Axial T2-weighted images in a 3-month-old boy showing bilateral small regions of occipital encephalomalacia characterized by linear hypointensity more evident on T2* imaging (image on the right), consistent with hemosiderin deposits (hemorrhagic encephalomalacia, arrowheads). *B*, Coronal inversion recovery images in a 5-month-old boy (patient 17) show signal abnormality and swelling of the left hippocampal formation (arrowheads). The examination was performed in concomitance with status epilepticus.



ON-LINE FIG 4. CT in a 4-month-old boy (patient 24) revealing numerous wormian bones (*small arrows*) that might simulate, on axial images, the presence of vault bone fractures. The central image is a magnified 3D reconstruction (volume-rendering technique) of the right pterion showing the wormian bones at the level of the sutures.