

Krabbe disease: An unusual presentation of optic nerve enlargement

**Authors:**

Maria Kaloianova¹
Jaishree Naidoo¹
Heather Thomson²
Louisa Bhengu³

Affiliations:

¹Department of Radiology,
University of the
Witwatersrand, South Africa

²Department of Paediatrics,
Charlotte Maxeke
Johannesburg Academic
Hospital, University of the
Witwatersrand, South Africa

³Department of Human
Genetics, National Health
Laboratory Services, School
of Pathology, University of
Witwatersrand, South Africa

Corresponding author:

Maria Kaloianova,
mkaloianova@gmail.com

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Krabbe disease is an autosomal recessive leukodystrophy that presents clinically with regression of milestones, excessive irritability and inconsolable crying. The pathologic basis of the disease is abnormal myelin metabolism resulting from a deficiency in the galactocerebrosidase enzyme with subsequent white matter destruction. Although optic atrophy is a classic presentation of Krabbe disease, we report on two patients who are biological brothers presenting with optic nerve enlargement in addition to other typical magnetic resonance imaging features of Krabbe disease, thereby confounding the initial diagnosis.

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