



## Abstracts

Articles appearing in the August 2020 issue

### Hydrocephalus and diffuse choroid plexus hyperplasia in primary ciliary dyskinesia-related MCIDAS mutation

**Objective** To report a neuroradiologic phenotype associated with reduced generation of multiple motile cilia (RGMC) and mutations in the *multicilin* gene. We hypothesize that the observed phenotype may reflect the emerging role that ependymal cilia play in regulating CSF production.

**Methods** Clinical and radiologic records were retrospectively reviewed for 7 consecutive patients diagnosed by the Leicester UK national primary ciliary dyskinesia (PCD) diagnostic laboratory.

**Results** On MRI scanning, all patients demonstrated hydrocephalus, choroid plexus hyperplasia (CPH), and arachnoid cysts. No patient had any sign of neurologic deficit. All patients had significant lung disease.

**Conclusions** We conclude that there is a high incidence of hydrocephalus, arachnoid cysts, and CPH in MCIDAS-associated RGMC. In all cases, the observed hydrocephalus seems arrested in childhood without progression or adverse neurologic sequelae. Our new observation of CPH, which is associated with CSF overproduction, is the first macroscopic evidence that ependymal cilia may be involved in the regulation of CSF production and flow. We suggest that brain imaging should be performed in all cases of RGMC and that a diagnosis of PCD or RGMC be strongly considered in patients with unexplained hydrocephalus and a lifelong “wet”-sounding cough.

[NPub.org/NG/9515a](https://pubmed.ncbi.nlm.nih.gov/39515a/)

### Delineating the phenotypic spectrum of sulfite oxidase and molybdenum cofactor deficiency

**Objective** To define the phenotypic spectrum of isolated sulfite oxidase (ISOD) and molybdenum cofactor deficiency (MoCD), aiming to promote timely diagnosis and assist in future clinical trial design.

**Methods** We analyzed clinical, radiographic, biochemical, and genetic data from 146 patients reported in the literature.

**Results** We stratified patients into 2 phenotypic subgroups based on clinical and radiographic characteristics. In the first (Class I), patients presented early in life (age 1–50 days) with acute onset of neurologic symptoms and development of diffuse brain injury with cystic leukomalacia. Patients in the second subgroup (Class II) presented later in life (age 30 days–23 years) with prominent movement abnormalities and selective injury of the basal ganglia and cerebellum. A significant difference in survival estimates correlated with milder disease severity among Class II patients. Substantial overlap in sulfur-containing metabolite levels prevented discrimination of subgroups based on diagnostic biomarkers, but genotype-phenotype correlations suggested that residual SUOX activity may contribute to milder phenotypes.

**Conclusions** Patients with SUOX and MoCD gravitate toward 1 of 2 distinct clinicoradiographic profiles. Patient stratification may help promote accurate diagnosis, prognostication, and aid in the design of future clinical trials.

[NPub.org/NG/9515b](https://pubmed.ncbi.nlm.nih.gov/39515b/)



## Most-Read Articles

As of August 11, 2020

### KCNQ2 encephalopathy: Features, mutational hot spots, and ezogabine treatment of 11 patients

J.J. Millichap, K.L. Park, T. Tsuchida, et al. 2016;2:e96. doi.org/10.1212/NXG.0000000000000096

### Mendelian randomization shows a causal effect of low vitamin D on multiple sclerosis risk

B. Rhead, M. Bäärnhielm, M. Gianfrancesco, et al. 2016;2:e97. doi.org/10.1212/NXG.0000000000000097

### CHCHD10 variant p.(Gly66Val) causes axonal Charcot-Marie-Tooth disease

M. Auranen, E. Ylikallio, M. Shcherbii, et al. 2015;1:e1. doi.org/10.1212/NXG.0000000000000003

### Homozygous deletion in MICU1 presenting with fatigue and lethargy in childhood

D. Lewis-Smith, K. J. Kamer, H. Griffin, et al. 2016;2:e59. doi.org/10.1212/NXG.0000000000000059

# Neurology®

What's happening in *Neurology*® *Genetics*  
*Neurology* 2020;95;684  
DOI 10.1212/WNL.0000000000010720

**This information is current as of October 12, 2020**

<b>Updated Information &amp; Services</b>	including high resolution figures, can be found at: <a href="http://n.neurology.org/content/95/15/684.full">http://n.neurology.org/content/95/15/684.full</a>
<b>Subspecialty Collections</b>	This article, along with others on similar topics, appears in the following collection(s): <b>All Genetics</b> <a href="http://n.neurology.org/cgi/collection/all_genetics">http://n.neurology.org/cgi/collection/all_genetics</a>
<b>Permissions &amp; Licensing</b>	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: <a href="http://www.neurology.org/about/about_the_journal#permissions">http://www.neurology.org/about/about_the_journal#permissions</a>
<b>Reprints</b>	Information about ordering reprints can be found online: <a href="http://n.neurology.org/subscribers/advertise">http://n.neurology.org/subscribers/advertise</a>

*Neurology*® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2020 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

