

ARTICLES
(/ARTICLES)

CASES (/CASES)

COURSES
(/COURSES)

PROFILE
(/USERS/YULIIA-
SOLODOVNIKOVA)



Introduction to oncology
staging CT
Sally Ayesa



(<https://radiopaedia.org/courses/featured-video>)

Myoclonic epilepsy with ragged red fibres (MERRF syndrome)



Case contributed by Yuliia Solodovnikova (/users/yuliia-solodovnikova)

Diagnosis almost certain



(/)

MENU

SEARCH

Edit case

Share (/cases/myoclonic-epilepsy-with-ragged-red-fibers-merrf-syndrome)

Add to

× Citation, DOI, disclosures and case data

Citation:

Solodovnikova Y, Myoclonic epilepsy with ragged red fibres (MERRF syndrome). Case study, Radiopaedia.org (Accessed on 13 Jun 2023)
<https://doi.org/10.53347/rID-155658>

DOI:

<https://doi.org/10.53347/rID-155658> (<https://doi.org/10.53347/rID-155658>)

Permalink:

<https://radiopaedia.org/cases/155658>
(<https://radiopaedia.org/cases/155658>)

rID:

155658

Disclosures:

At the time the case was submitted for publication Yuliia Solodovnikova had no financial relationships to ineligible companies (</articles/disclosures>) to disclose.



View Yuliia Solodovnikova's current disclosures (</users/yuliia-solodovnikova>)

Case created:

1 Nov 2022

Case published:

27 Feb 2023

Revisions:

14 times, by 4 contributors - see full revision history and disclosures (</cases/myoclonic-epilepsy-with-ragged-red-fibers-merrf-syndrome/revisions>)

Systems:

Central Nervous System (</articles/section/all/central-nervous-system>)

Case of the day:

07 Jun 2023

Institution:

ONMedU

Quiz mode:

Excluded

Presentation

Manifested in early teens with epileptic seizures, general weakness, progressive cognitive decline, emotional lability, shaky writing, and walking. Tonic-clonic seizures were successfully treated with a

combination of two antiepileptic drugs, but the eradication of myoclonic status wasn't achieved. The patient also has hypothyroidism and cardiomyopathy.



Patient Data

Age: 20 years

Gender: Female

MRI

(/cases/155658/studies/128218)



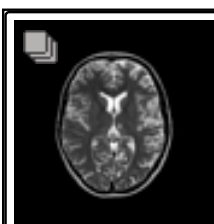
Axial FLAIR

(/images/59999
645?
case_id=155658
)



Sagittal FLAIR

(/images/59999
474?
case_id=155658
)



Axial T2

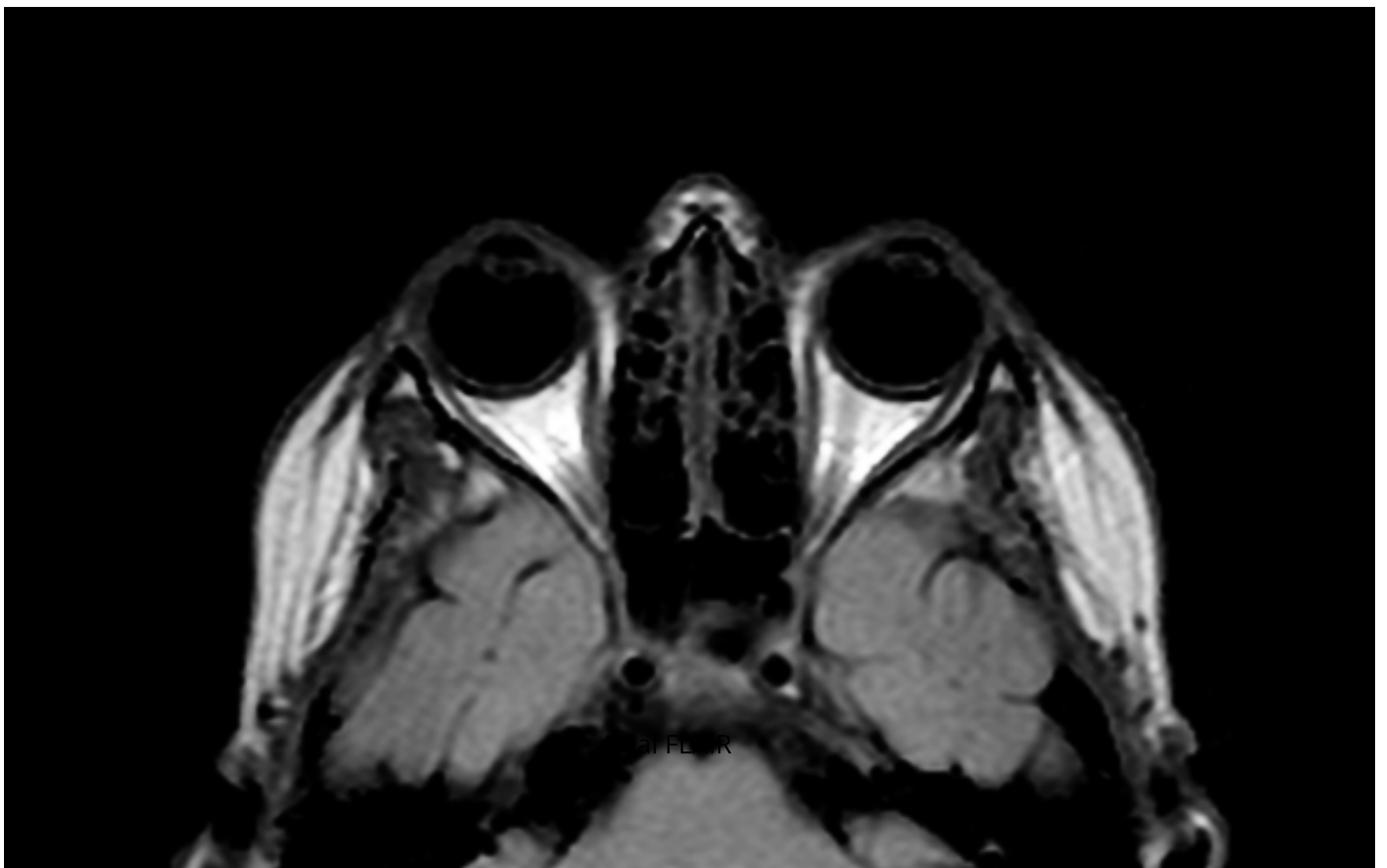
(/images/59999
628?
case_id=155658
)

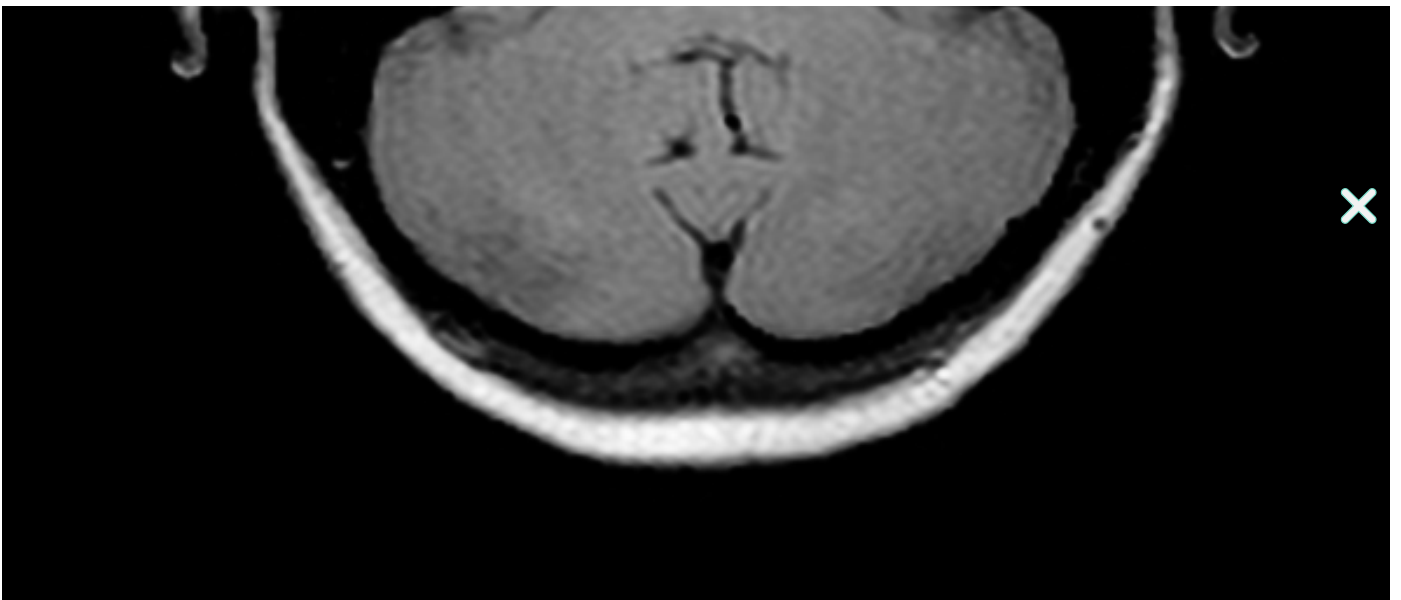


Axial T1 C+

(/images/59999
695?
case_id=155658
)

MRI





patient might have mitochondrial pathology. Genetic analysis has shown the mutation in the MT-ND5 gene. The latter is characteristic of MERRF syndrome (<https://radiopaedia.org/articles/myoclonic-epilepsy-with-red-ragged-fibres-merrf-1>), MELAS (<https://radiopaedia.org/articles/mitochondrial-encephalomyopathy-with-lactic-acidosis-and-stroke-like-episodes-melas>), and Leigh syndrome (<https://radiopaedia.org/articles/leigh-syndrome-3>), of which MERRF syndrome was the most likely diagnosis, judging by the clinical picture. Unfortunately, the patient refused to perform a muscle biopsy to confirm the diagnosis.


Case Discussion

This case shows both the radiological and clinical picture of MERRF syndrome (</articles/myoclonic-epilepsy-with-red-ragged-fibres-merrf-1>). Primary mitochondrial disorders (</articles/primary-mitochondrial-disorders>) are especially hard to diagnose properly, so a thorough differential diagnosis is required, relying on both on MRI and genetic analysis.

+ **1 public playlist includes this case**

× **Related Radiopaedia articles**

- Leigh syndrome (</articles/leigh-syndrome-3>)

- [Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes \(MELAS\) \(/articles/mitochondrial-encephalomyopathy-with-lactic-acidosis-and-stroke-like-episodes-melas\)](/articles/mitochondrial-encephalomyopathy-with-lactic-acidosis-and-stroke-like-episodes-melas) 
 - [Myoclonic epilepsy with red ragged fibres \(MERRF\) \(/articles/myoclonic-epilepsy-with-red-ragged-fibres-merrf-1\)](/articles/myoclonic-epilepsy-with-red-ragged-fibres-merrf-1)
 - [Primary mitochondrial disorders \(/articles/primary-mitochondrial-disorders\)](/articles/primary-mitochondrial-disorders)
-

× **Promoted articles** (advertising)

Video Series: Anemia in Chronic Kidney Disease in Primary Care
Stephen Brunton et al, Clinical Diabetes

Clinical Diabetes Video Series: Anemia in CKD in Primary Care
Stephen Brunton et al, Clinical Diabetes

Video Series: Anemia in CKD in Primary Care
Stephen Brunton et al, Clinical Diabetes

Using Virtual Reality Head-Mounted Displays to Assess Skills in
Emergency Medicine: Validity Study
Marie Høxbro Knudsen et al., J Med Internet Res, 2023

Clinical Diabetes CME: Optimizing the Use of GLP-1RAs in Type
2 Diabetes
John Anderson et al., Clinical Diabetes

Podcast: Low and No Calorie Sweeteners, from Diabetes Core
Update
Diabetes Core Update, 2023

I consent to the use of Google Analytics and related cookies across the TrendMD network (widget, website, blog). [Learn more \(https://www.trendmd.com/google-analytics#\)](https://www.trendmd.com/google-analytics#)



Yes

No

Case completeness

80% complete. Why not add:

- Diagnostic certainty
- Patient's presentation
- Patient data
- Study findings
- Case discussion
- MRI image sequences
- Modality
- Related articles
- A couple of questions and answers

How to use cases

You can use Radiopaedia cases in a variety of ways to help you learn and teach.

- Add cases to playlists (/articles/playlists)
- Share cases with the diagnosis hidden (/articles/sharing-cases-and-playlists)

- Use images in presentations (</articles/using-and-attributing-images-from-radiopaedia>)
- Use them in multiple choice question (</articles/image-interpretation-questions>)



Creating your own cases is easy.

- Case creation learning pathway (</courses/help-creating-cases>)

ADVERTISEMENT: Supporters see fewer/no ads

Articles (</articles/section/all/all>)

By Section:

Anatomy

(</articles/section/anatomy/all>)

Approach

(</articles/section/approach/all>)

Artificial Intelligence

(</articles/section/artificial->

Cases (</cases>)

Breast (</cases/system/breast>)

Cardiac (</cases/system/cardiac>)

Central Nervous System

(</cases/system/central-nervous-system>)


Chest (</cases/system/chest>)

Forensic (</cases/system/forensic>)

intelligence/all)
Classifications
(/articles/section/classifications/all)
Gamuts
(/articles/section/gamuts/all)
Imaging Technology
(/articles/section/physics/all)
Interventional Radiology
(/articles/section/interventional-radiology/all)
Mnemonics
(/articles/section/mnemonics/all)
Pathology
(/articles/section/pathology/all)
Radiography
(/articles/section/radiography/all)
Signs (/articles/section/signs/all)
Staging
(/articles/section/staging/all)
Syndromes
(/articles/section/syndromes/all)

By System:

Breast (/articles/section/all/breast)
Cardiac
(/articles/section/all/cardiac)
Central Nervous System
(/articles/section/all/central-nervous-system)
Chest (/articles/section/all/chest)

Gastrointestinal
(/cases/system/gastrointestinal)
Gynaecology
(/cases/system/gynaecology) 
Haematology
(/cases/system/haematology)
Head & Neck
(/cases/system/head-neck)
Hepatobiliary
(/cases/system/hepatobiliary)
Interventional
(/cases/system/interventional)
Musculoskeletal
(/cases/system/musculoskeletal)
Obstetrics
(/cases/system/obstetrics)
Oncology
(/cases/system/oncology)
Paediatrics
(/cases/system/paediatrics)
Spine (/cases/system/spine)
Trauma (/cases/system/trauma)
Urogenital
(/cases/system/urogenital)
Vascular (/cases/system/vascular)
Not Applicable
(/cases/system/not-applicable)

Radiopaedia.org (/)

ABOUT (/ABOUT)

Forensic
(/articles/section/all/forensic)

Gastrointestinal
(/articles/section/all/gastrointestinal)

Gynaecology
(/articles/section/all/gynaecology)

Haematology
(/articles/section/all/haematology)

Head & Neck
(/articles/section/all/head-neck)

Hepatobiliary
(/articles/section/all/hepatobiliary)

Interventional
(/articles/section/all/interventional)

Musculoskeletal
(/articles/section/all/musculoskeletal)

Obstetrics
(/articles/section/all/obstetrics)

Oncology
(/articles/section/all/oncology)

Paediatrics
(/articles/section/all/paediatrics)

Spine (/articles/section/all/spine)

Trauma
(/articles/section/all/trauma)

Urogenital
(/articles/section/all/urogenital)

Vascular
(/articles/section/all/vascular)

BLOG (/BLOG)

FEATURE SPONSOR (/FEATURE-SPONSOR)

DONATE (/SUPPORTERS)



Editors (/editors)

Expert advisers (/expert-advisers)

Help (/articles/style-guide-and-help)



Facebook

(<https://www.facebook.com/Radiopaedia>)



Twitter

(<https://twitter.com/Radiopaedia>)



Newsletter

(<http://eepurl.com/ltOWf>)

[Contact Us \(/contact\)](/contact)

[Terms of Use \(/terms\)](/terms)

[Privacy Policy \(/terms#privacy-policy\)](/terms#privacy-policy)

[Licensing \(/licence\)](/licence)



[Sponsorship \(/sponsorship\)](/sponsorship)

[Developers \(/developers\)](/developers)

© 2005–2023 Radiopaedia.org