

# Orbital involvement in Erdheim–Chester Disease

Presenter: Fatemeh Dehghani Firouzabadi, MD

Mahshid Golagha, Aryan Zahergivar, Fahimul Huda, Jhanavi  
Rao, Nadia Biassou, Rahul H. Dave, Juvianee I. Estrada-Veras,  
Kevin O'Brien, Ashkan A. Malayeri

Department of Radiology and Imaging Sciences  
National Institutes of Health



# Disclosure:

We have no financial disclosure or conflicts of interest with the presented materials in this presentation.

# Purpose:

- ✓ To assess the orbital involvement in ECD patients.
- ✓ To evaluate associations between radiologic orbital findings with the BRAFV600E mutation.

# Material & Methods:

- ✓ MRI studies were examined by two radiologists, and any disagreements were settled by a third reader.
- ✓ Reviewing radiologists were only aware of pathology-confirmed diagnosis and were not aware of any genetic (BRAFV600E status) or clinical information.
- ✓ IRB approved study on 54 subjects with biopsy-confirmed diagnosis of ECD.

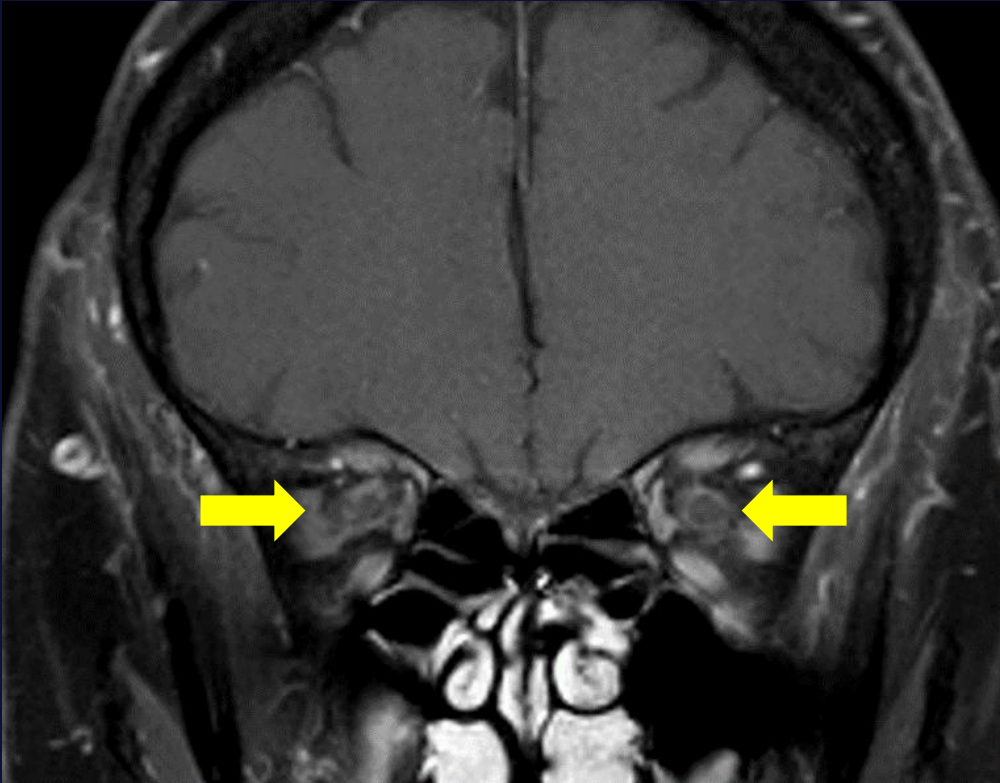
# Results:

Orbital Involvement	Number of Patients (54)	Percentage (%)
<b>Bilateral rectus muscle</b>	3	5
Bilateral lateral rectus muscle	2	4
Bilateral superior rectus muscle	1	2
Bilateral inferior rectus muscle	1	2
Bilateral medial rectus muscle	1	2
<b>Bilateral Retroorbital fat</b>	9	18
Left Retroorbital fat	2	4
<b>Optic nerve(leptomeningeal)</b>	1	2

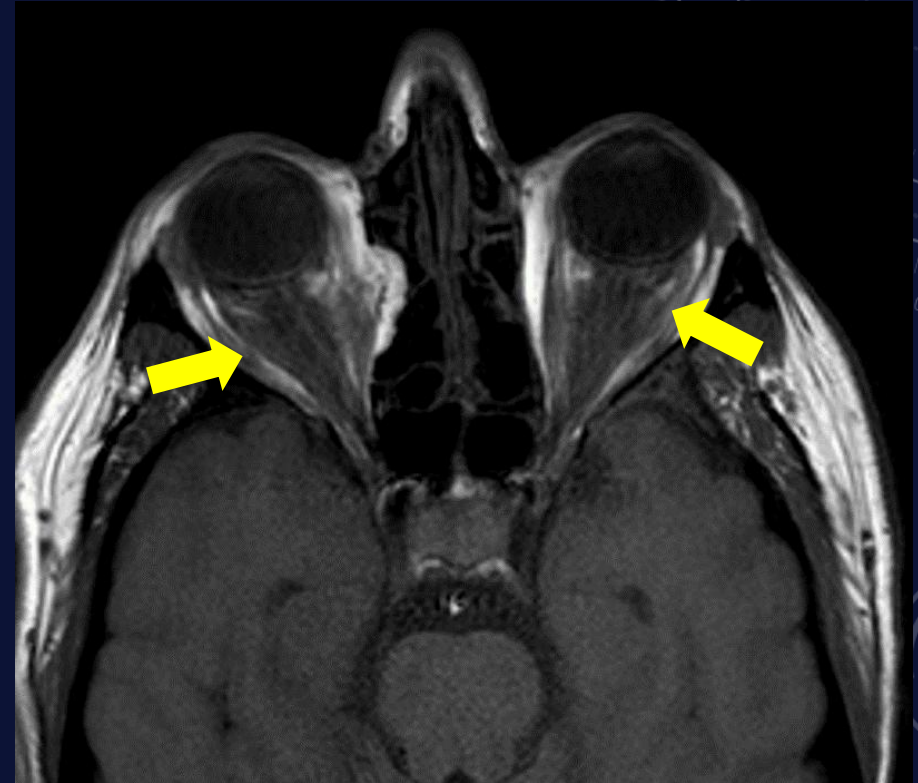
# Results:

- One patient with a BRAFV600E mutation showed optic nerve (leptomeningeal) involvement.
- Two patients with this mutation had left retroorbital fat involvement. However, BRAFV600E mutation wasn't significantly associated with bilateral rectus muscle or bilateral retroorbital fat involvement.

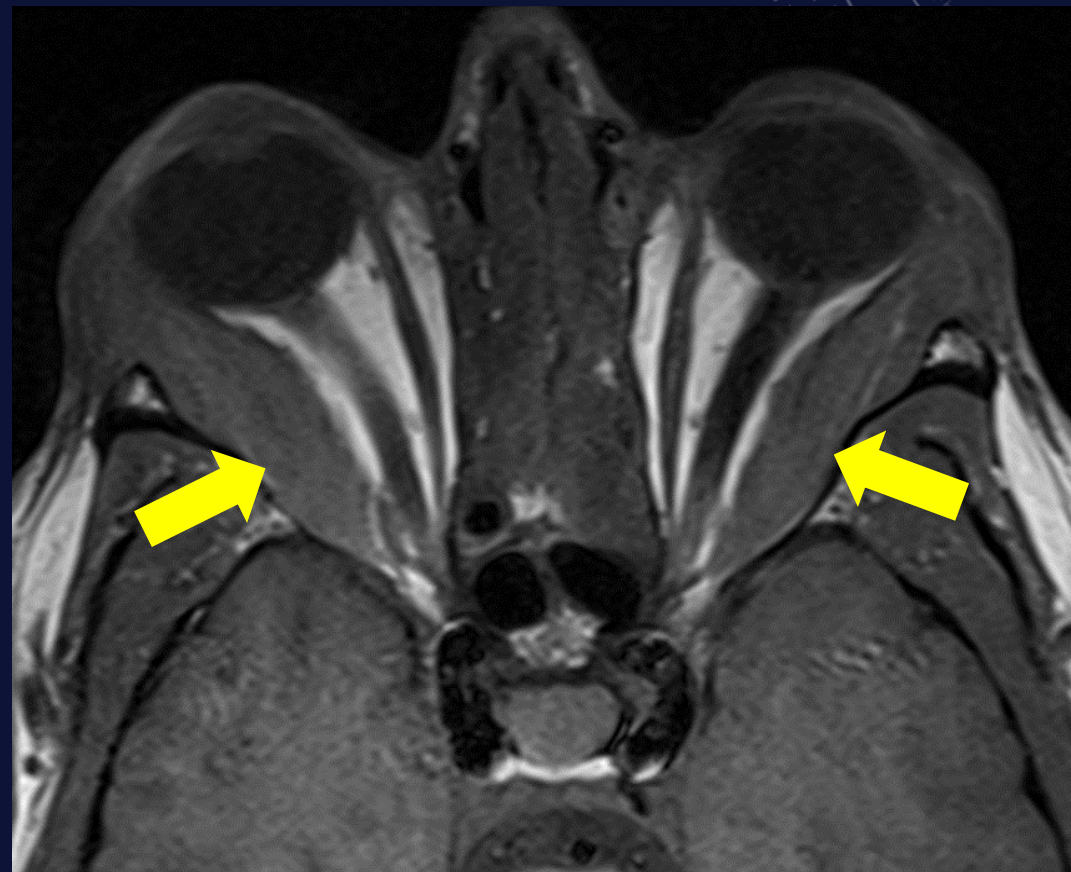
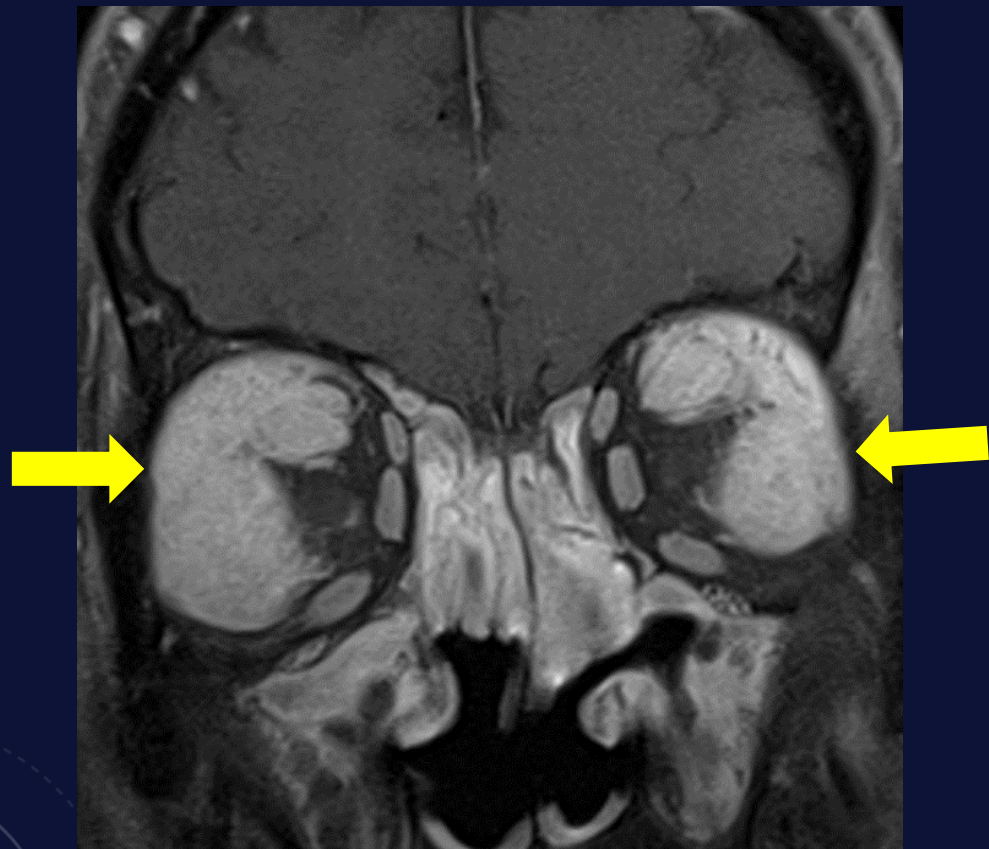
## Optic nerve involvement



## Bilateral retroorbital fat infiltration



# Bilateral superior and lateral rectus muscles with medial displacement of optic nerve



# Conclusion:

- Largest single-center study, which included 54 patients with ECD, showed orbital imaging findings including optic nerve, rectus muscle, and retroorbital fat involvement.
- Our findings showed no significant association between BRAFV600E mutation and bilateral rectus muscle or bilateral retroorbital fat involvement while one case with a BRAFV600E mutation showed optic nerve (leptomeningeal) involvement

# Thank you for your attention!



Mina.df20@gmail.com



@FirouzabadiMina

