Central Nervous System Involvement in Erdheim-Chester Disease

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Disclosure:

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

Purpose:

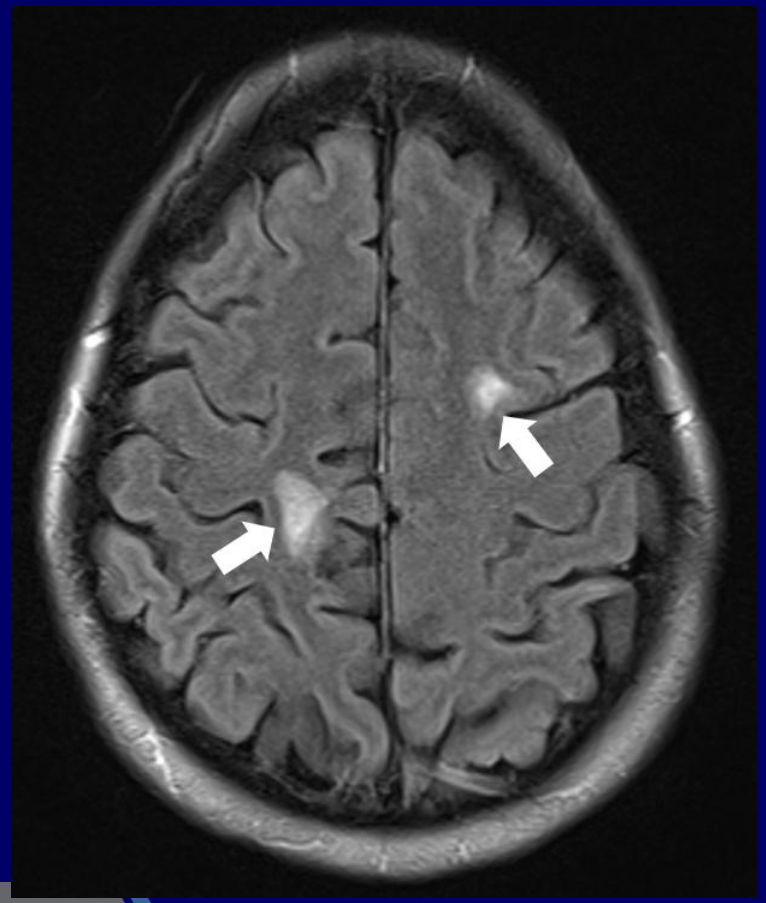
- ✓ Assessing distribution and patterns of CNS involvement in ECD.
- ✓ Evaluating potential association between presence of the BRAFV600E mutation and radiological findings in CNS.

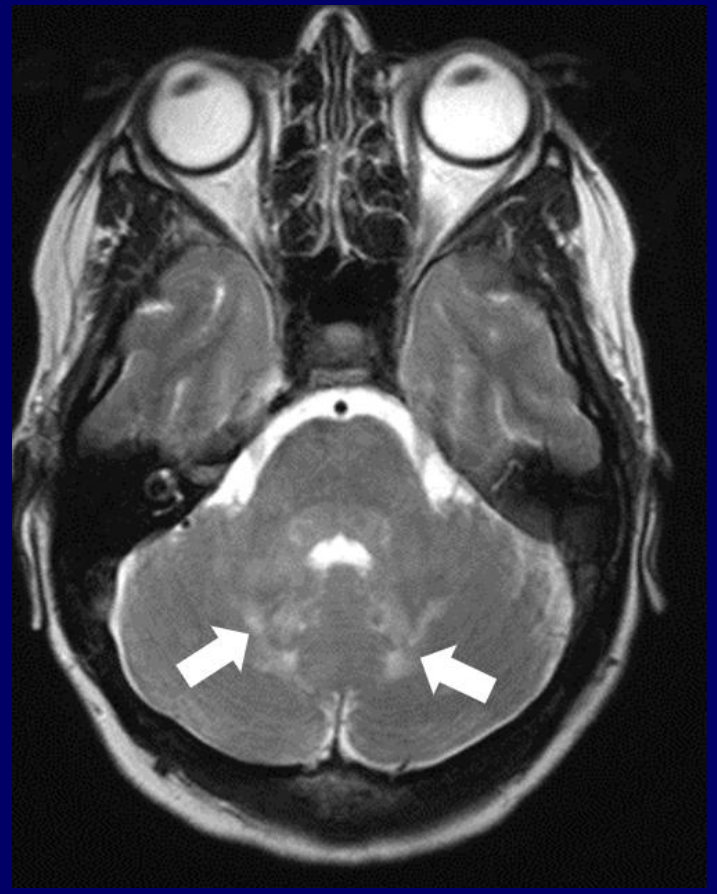
Material & Methods: Patient selection

- ✓ IRB approved study on 58 subjects with biopsy-confirmed diagnosis of ECD.
- ✓ Two radiologists independently reviewed the images.
- ✓ Discrepancies between the two radiologists were resolved through third neuroradiologist.

Cortical lesions: 46.6%
Bilateral cortical lesions: 32.8%
Frontal lobe lesions: 41%
Over 90% of lesions were well-demarcated

Cerebellar lesions: 25.9%
Dentate nucleus lesions: 15%





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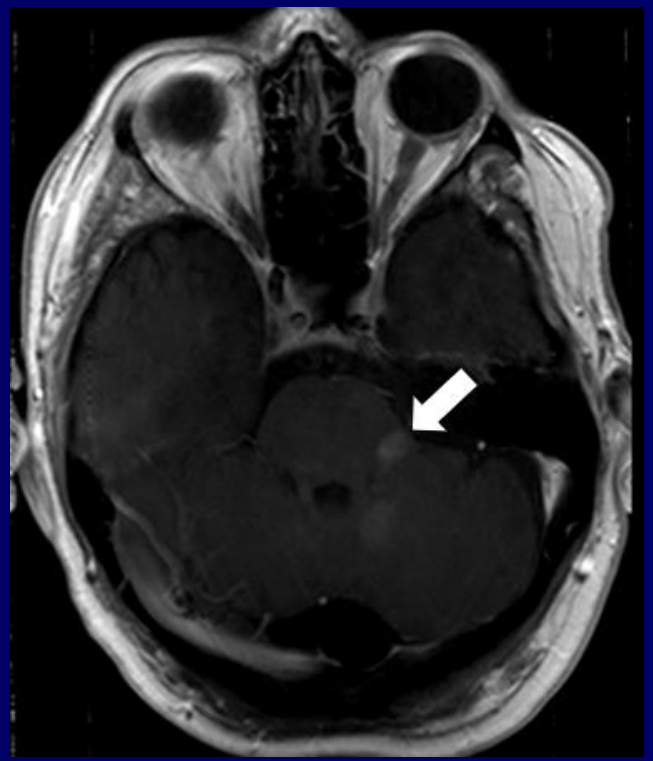
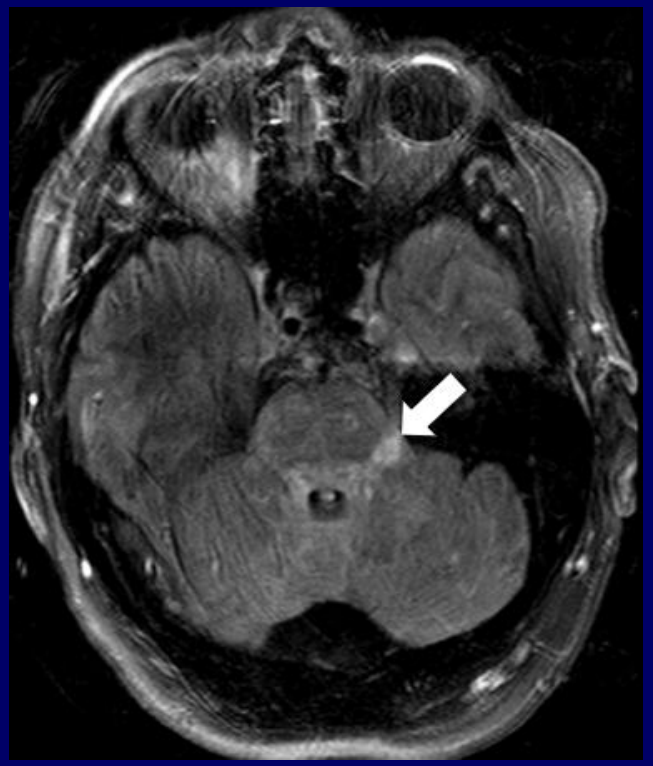
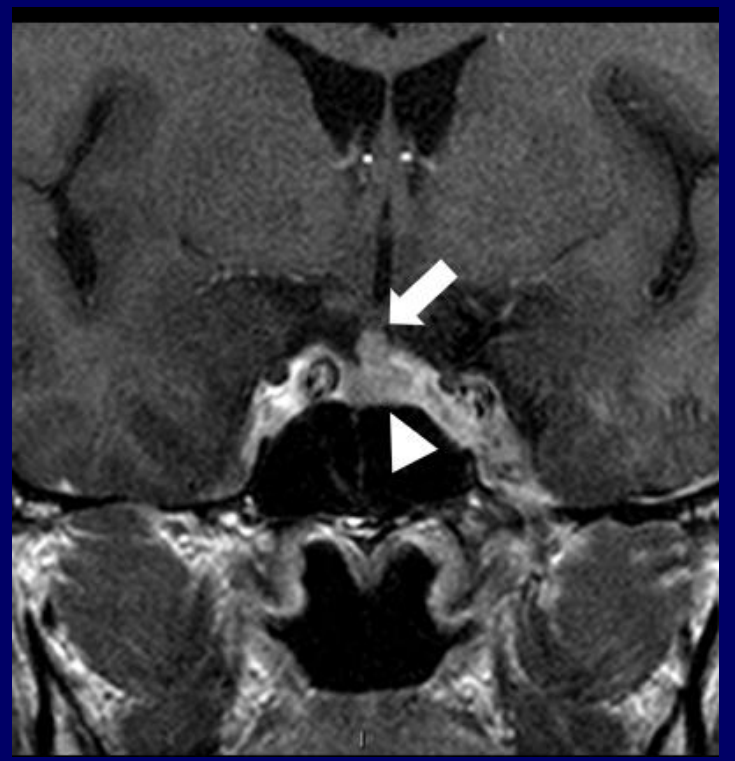
ERDHEIM-CHESTER DISEASE GLOBAL ALLIANCE

- ✓ Brainstem lesions: 29.3%
- ✓ Pons lesions: 26%



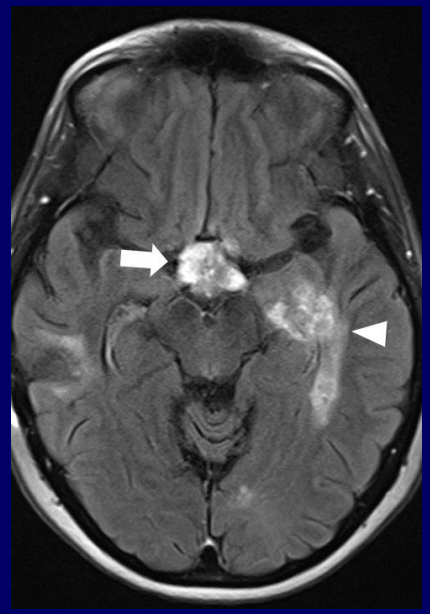


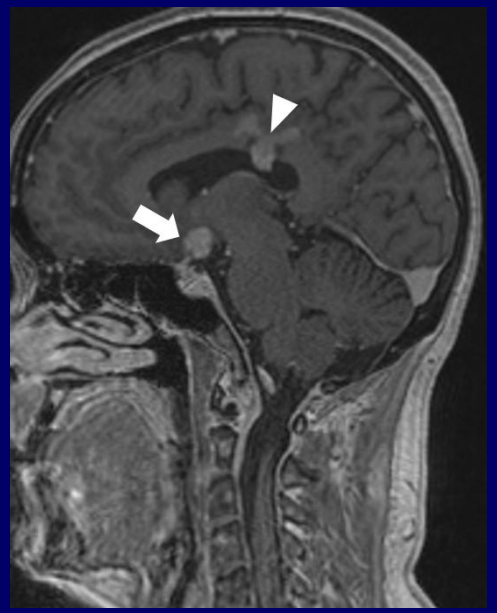


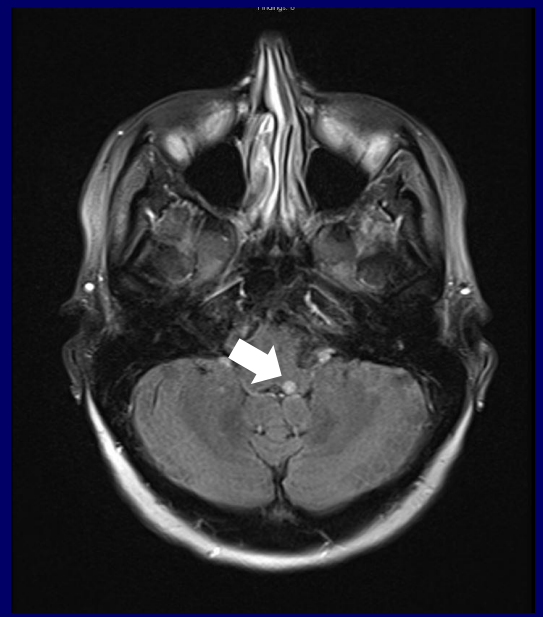
Pituitary lesions: 17.2%
Infundibular stalk lesions:14%

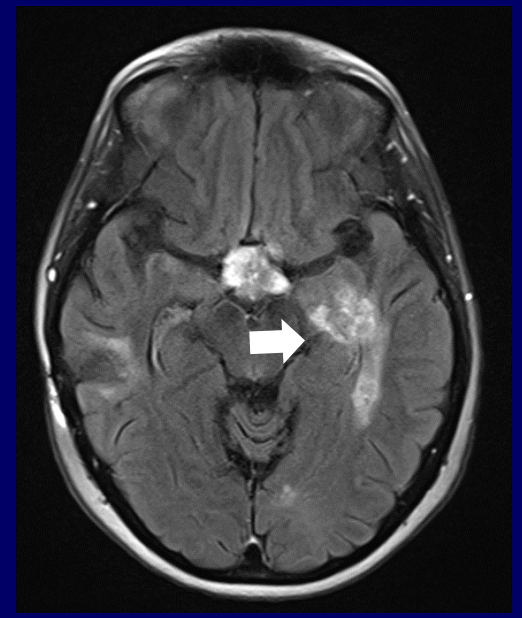
Basal Ganglia lesions: 6.9%
Putamen lesions: 5%

Amygdala lesions: 3%
Thalamic lesions: 3%









Results: Associations with BRAFV600E

- ✓ BRAFV600E mutation was significantly associated with cerebellar and bilateral brain stem lesions.
- ✓ A trend toward significance was noted for cerebral atrophy.

Conclusion

The results highlight significant radiologic CNS involvement in ECD and suggest potential associations with BRAFV600E mutation that could have diagnostic, prognostic, and therapeutic implications.


Thank you for your attention
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