

Overview of Histiocytic Neoplasms

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10th Annual ECDGA Patient & Family Gathering
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O'NEAL COMPREHENSIVE
CANCER CENTER
UAB MEDICINE.

Conflicts of Interest

- Royalties from UpToDate
- Consulting fees from Recordati
- Advisory board: Opna Bio, SeaGen, SOBI

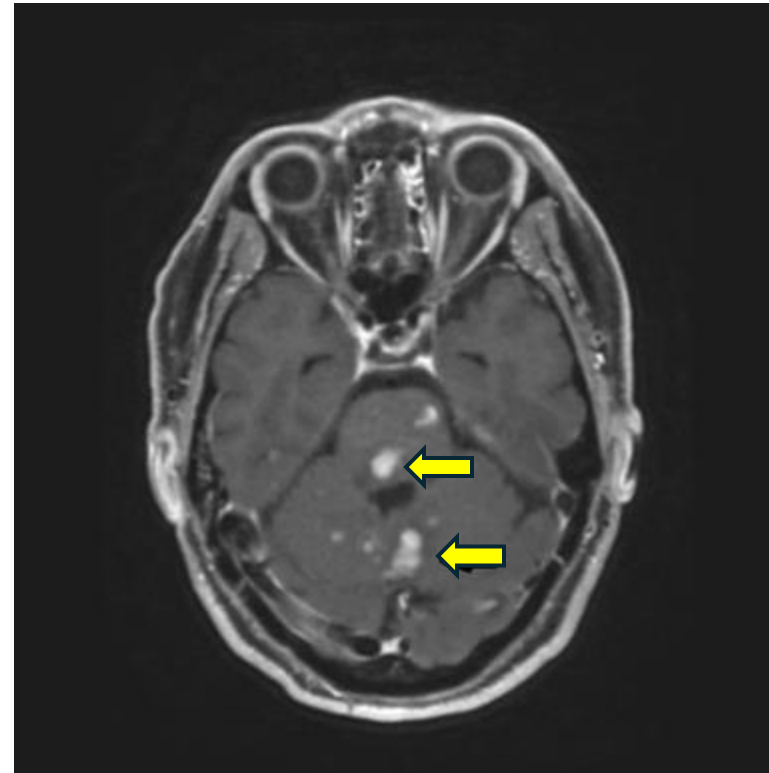
Case from the clinic

55 y/o male

1990s
Left thigh tingling and weakness

2008
Right hand tingling
MRI brain lesions
Diagnosed with multiple sclerosis

2017
Difficulty talking and walking
Multiple treatments for MS
Brain biopsy- inflammation



Histiocytic Neoplasms

- Rare (3-5 cases/million/year)
- Growth of abnormal blood cells (histiocytes) in organs
- Can affect any organ system

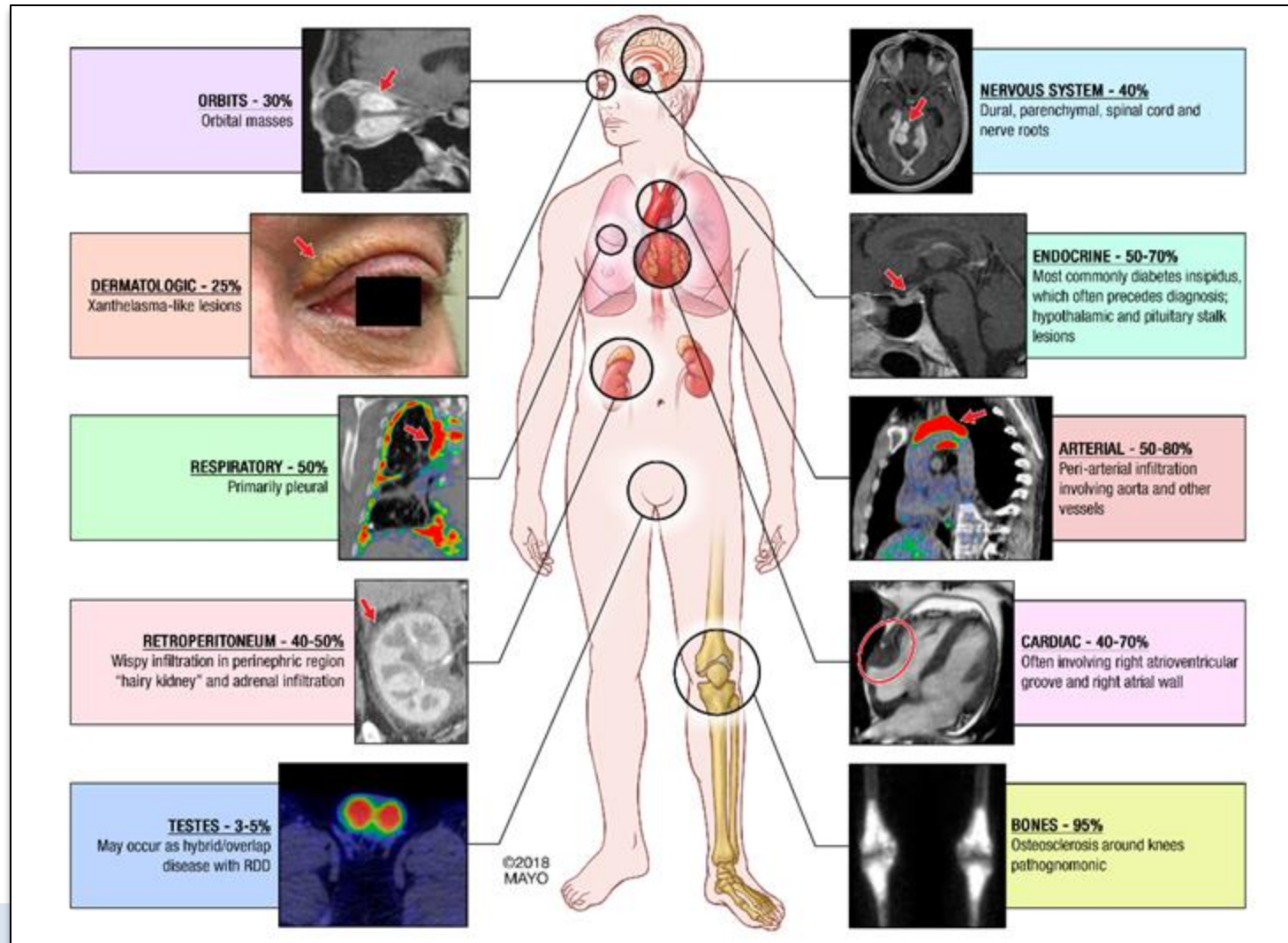
Chronic (slow growing)

- Erdheim-Chester disease
- Xanthogranuloma family disorders
- Rosai-Dorfman disease
- Langerhans cell histiocytosis

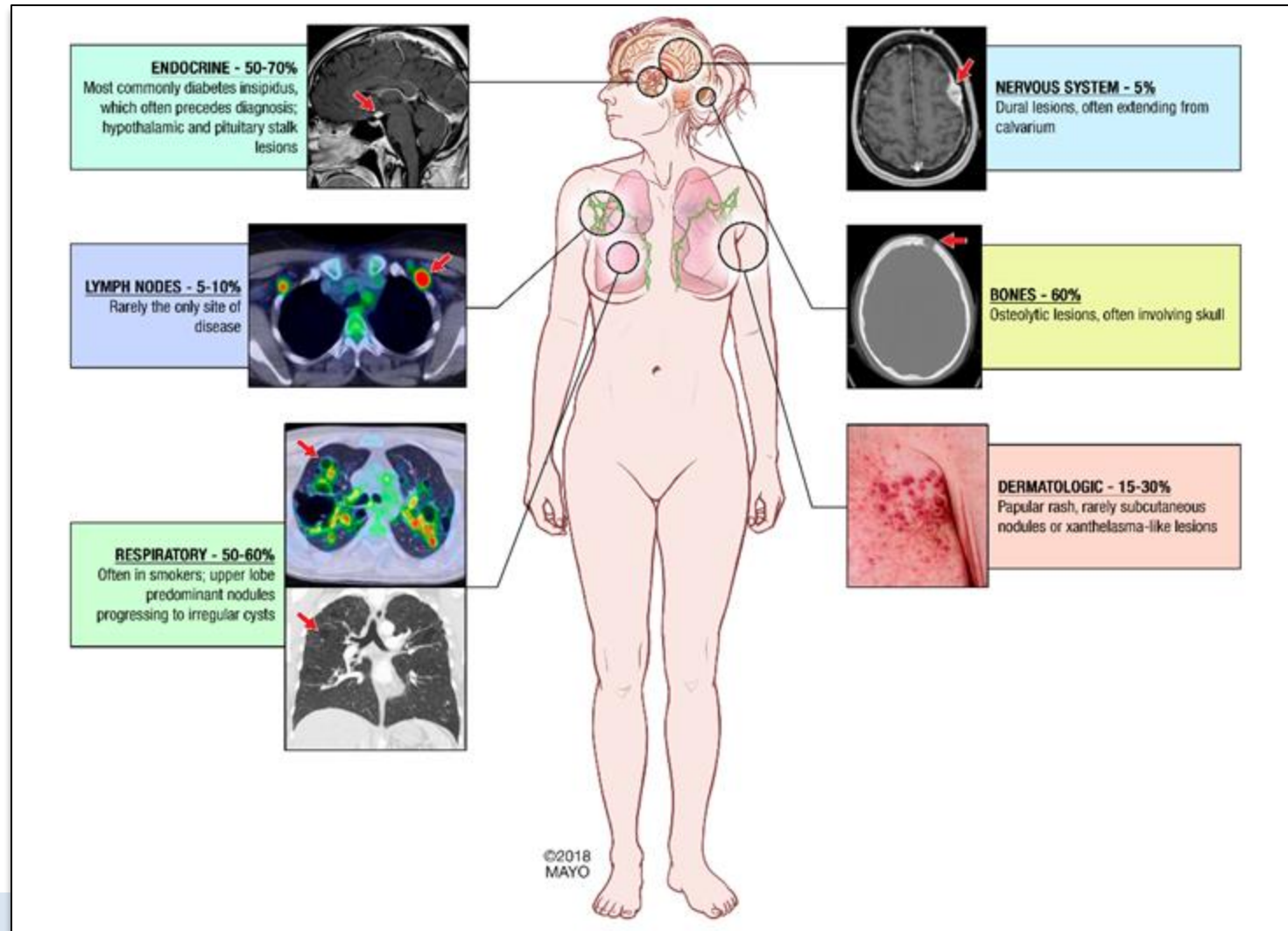
Aggressive (fast growing)

- Histiocytic sarcoma
- Langerhans cell sarcoma
- Interdigitating dendritic cell sarcoma

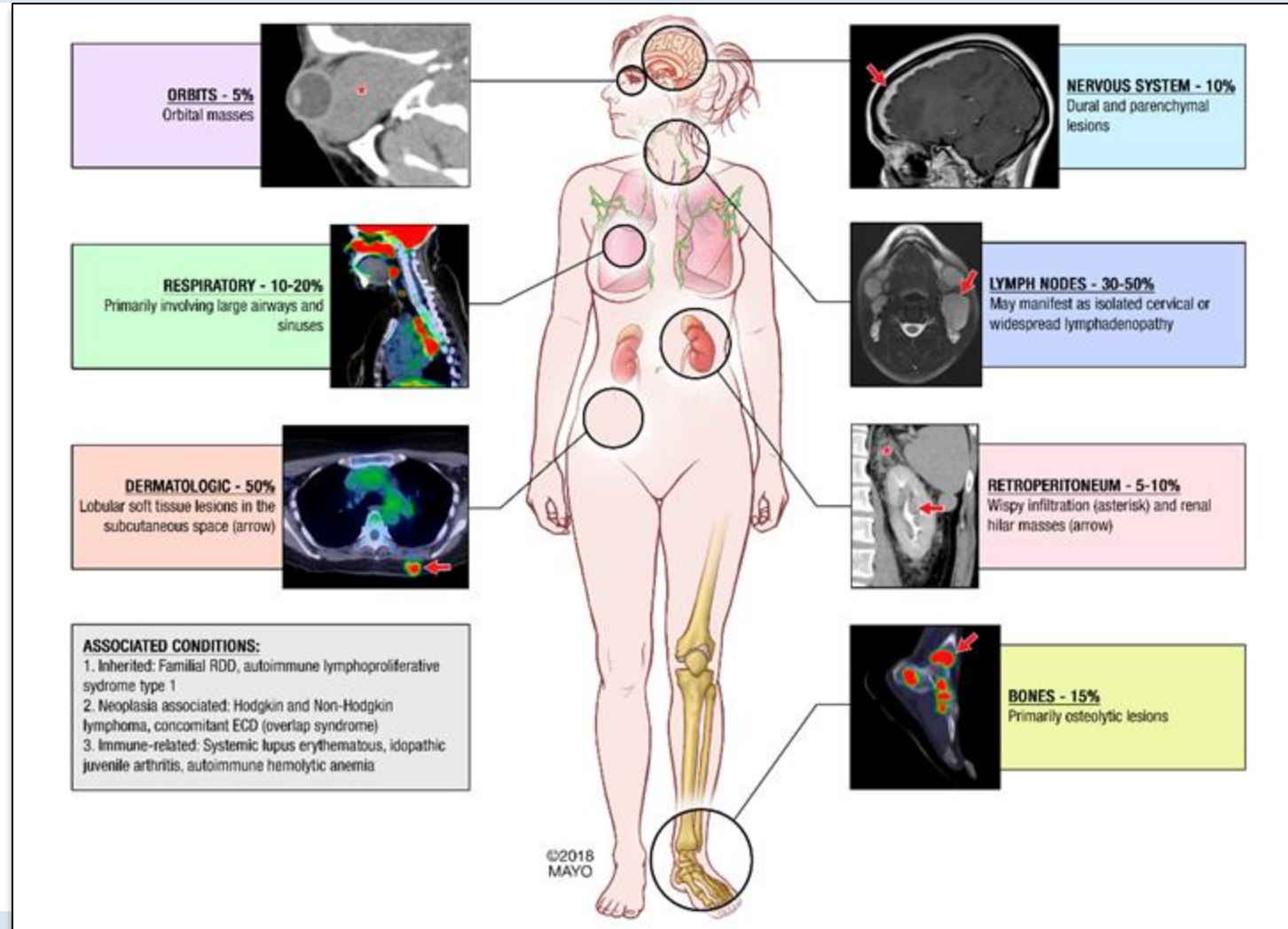
Manifestations of ECD



Manifestations of LCH

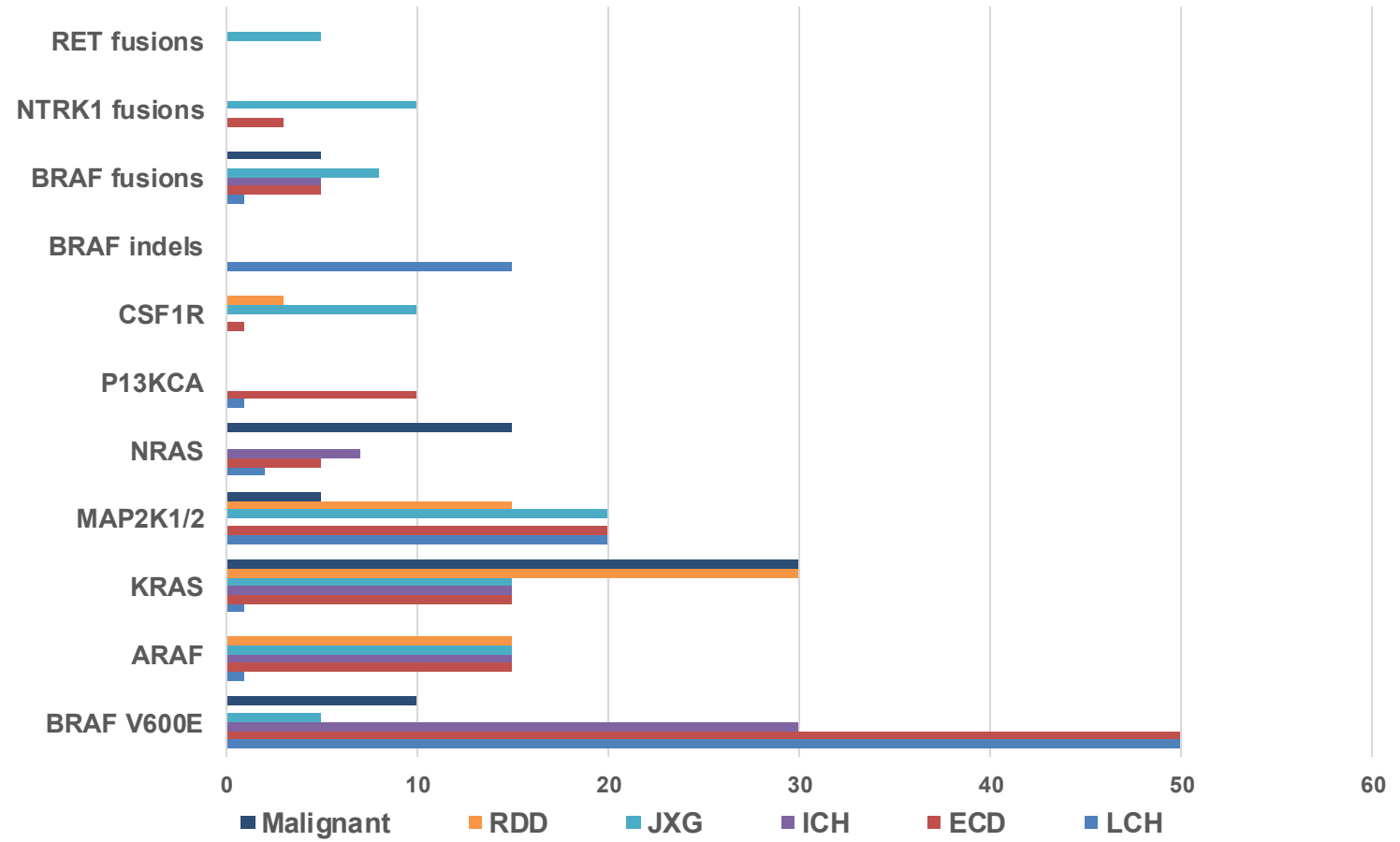
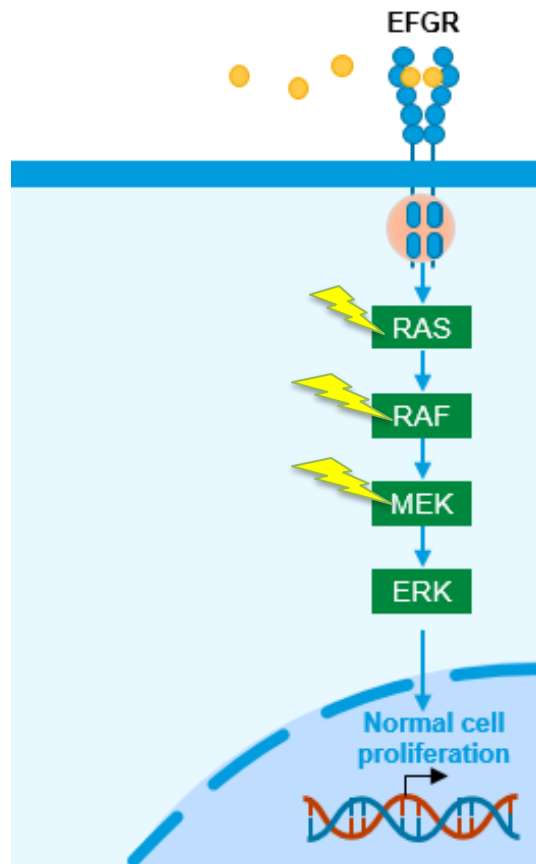


Manifestations of RDD

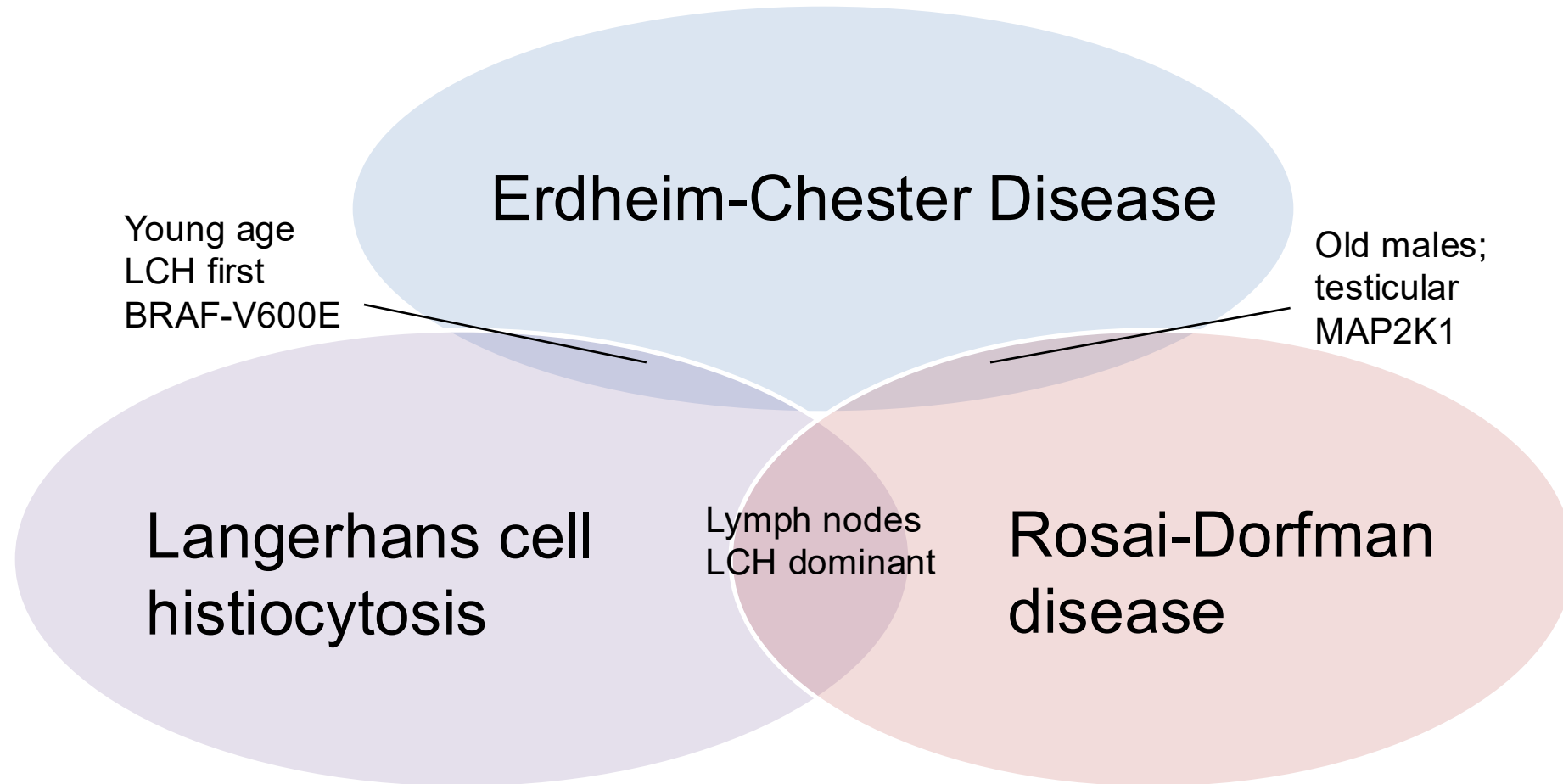


MAPK pathway mutations in histiocytosis

Normal cell growth



Overlap or mixed histiocytic neoplasms



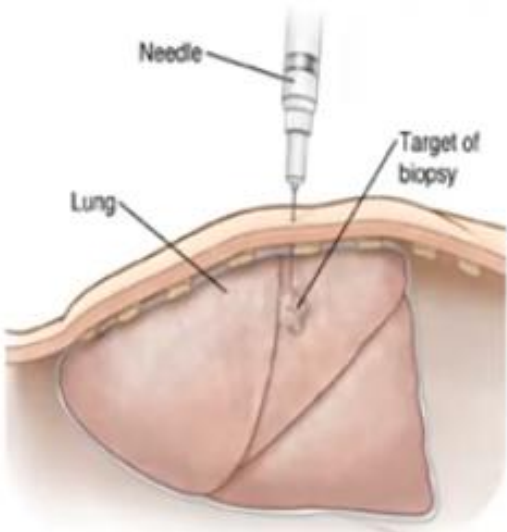
Hervier B, et al. Blood. 2014 Aug 14;124(7):1119-26

O'Malley DP et al. Mod Pathol. 2010;23(12):1616-1623.

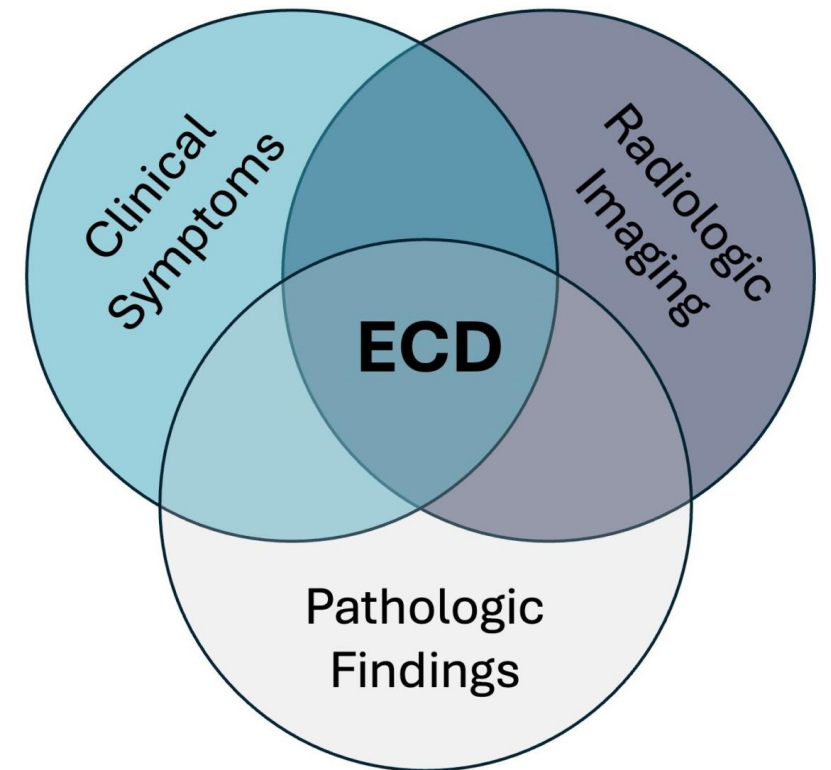
Razanamahery J,...Goyal G, et al. Haematologica. 2020 Jan;105(1):e5-e8

How to diagnose histiocytosis?

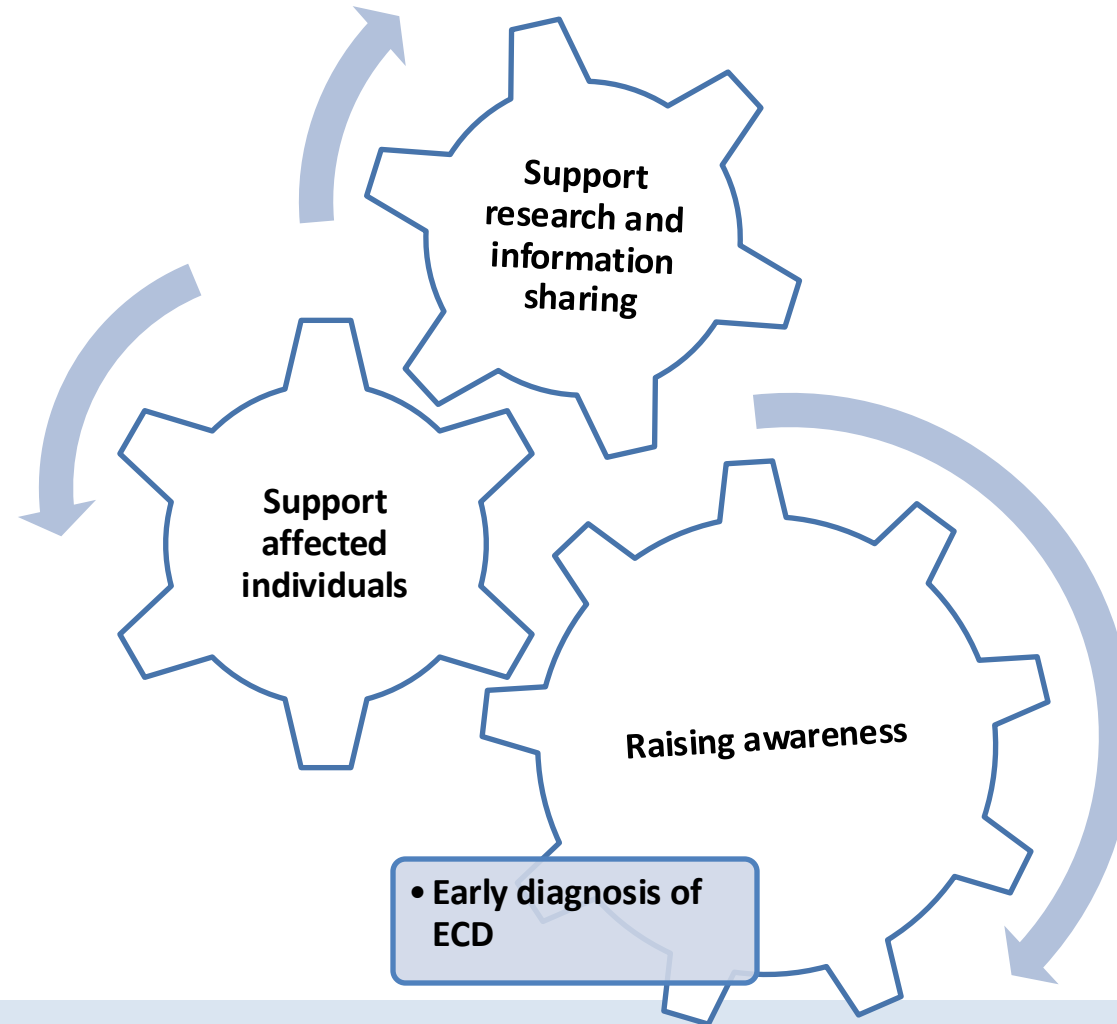
Most histiocytic neoplasms



Erdheim-Chester Disease
Diagnosis Requirements



ECDGA Mission



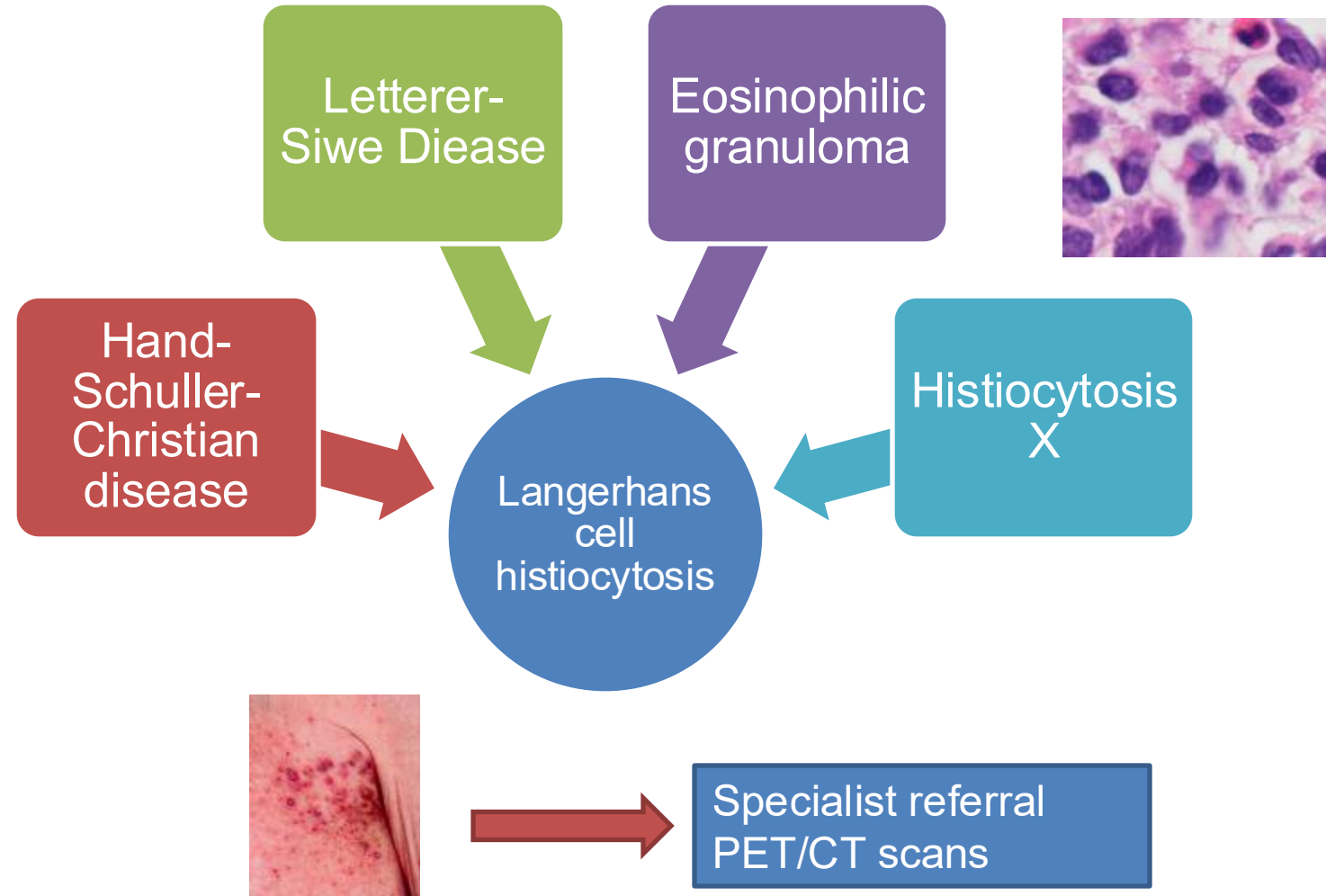
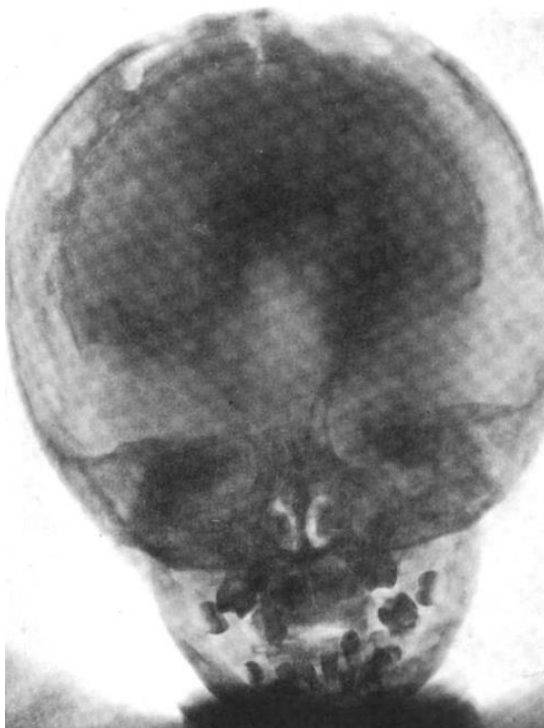
History Archives: LCH

DEFECTS IN MEMBRANOUS BONES, EXOPHTHALMOS, AND DIABETES INSIPIDUS

AN UNUSUAL SYNDROME OF DYSPITUITARISM; A CLINICAL STUDY

By HENRY A. CHRISTIAN, M.D., BOSTON, MASS.

(From the Medical Clinic of the Peter Bent Brigham Hospital, Boston, Mass.)

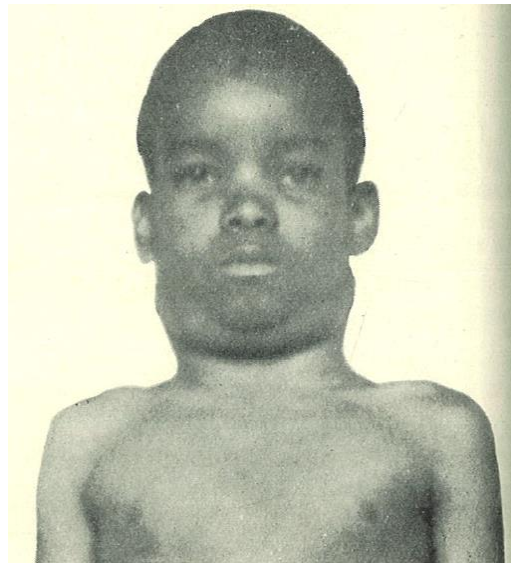


History Archives: RDD

Sinus Histiocytosis With Massive Lymphadenopathy

A Newly Recognized Benign Clinicopathological Entity

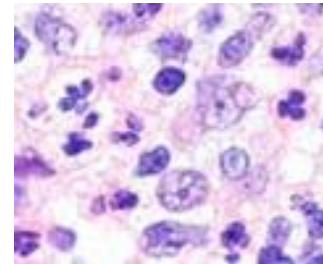
Juan Rosai, MD, and Ronald F. Dorfman, MBBCh, St. Louis



Adenitis with lipid
excess

Sinus histiocytosis
with massive
lymphadenopathy

Rosai-
Dorfman
disease



Specialist referral
PET/CT scans

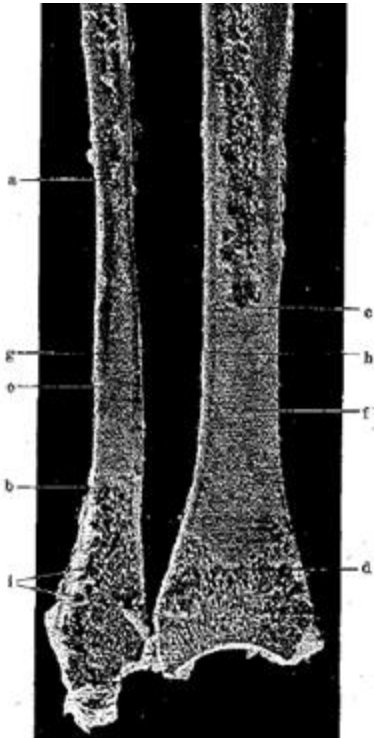
History Archives: ECD

Über Lipoidgranulomatose¹.

Von
Dr. William Chester, New-York.

Mit 9 Abbildungen im Text.

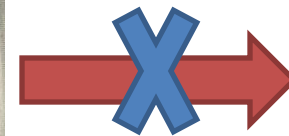
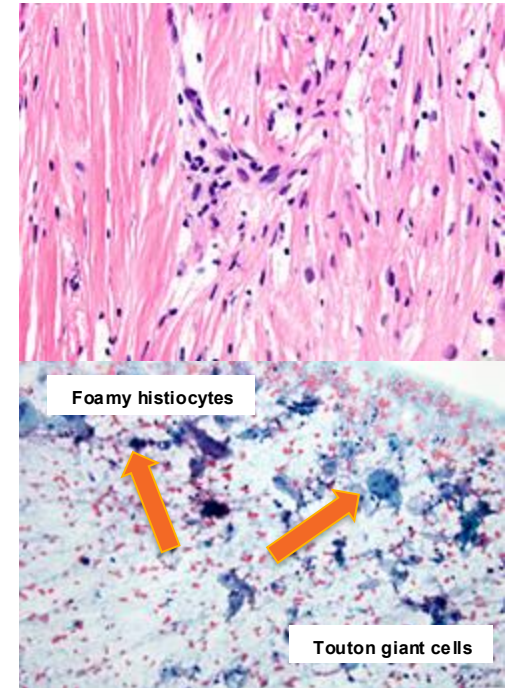
(Eingegangen am 29. Mai 1930.)



Average 6 organ systems affected

BRAF mutation 65-70%

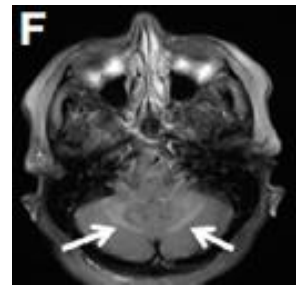
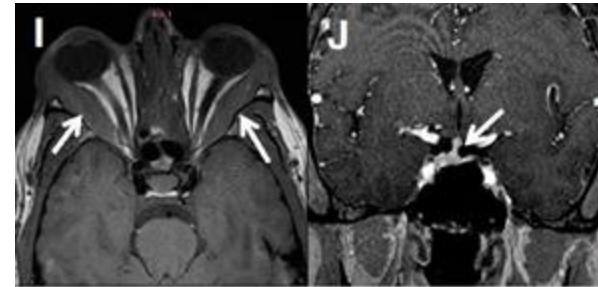
Xanthogranuloma



Specialist referral
PET/CT scans

Can we achieve early ECD diagnosis?

What does 'early ECD' look like?



"Hairy kidney"



Right atrial mass



"Coated aorta"



ECD without bone involvement
Average 3 organs involved, BRAF ~15%

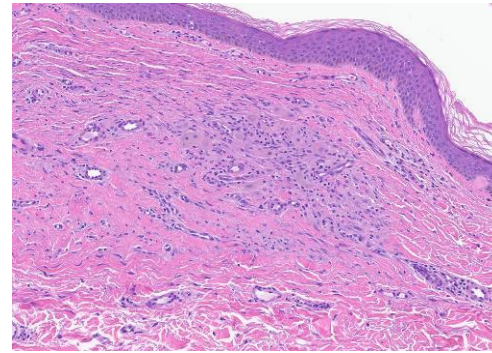
Case Conclusion

55 y/o male
Difficulty talking and walking

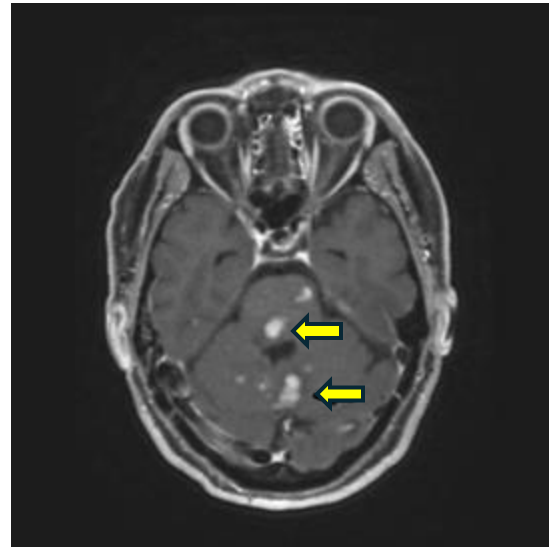
2023
Worsening weakness
Went to another neurologist

2024
Suspicion of ECD
Skin exam showed thigh lesion
Biopsy: Xanthogranuloma

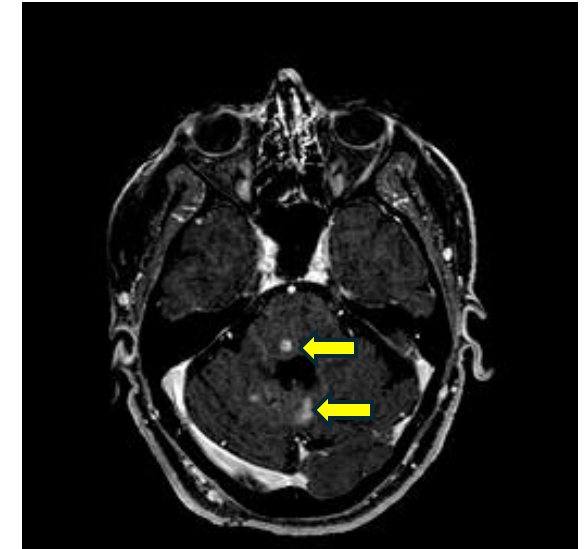
2024
Diagnosed with ECD
Started cobimetinib
Partial response



ECD



Cobimetinib



UAB Multidisciplinary Histiocytosis Program



Hematopathology
• Aishwarya Ravindran



Hematopathology
• Diana Morlote



Hematology
• Gaurav Goyal



Hematology
• Aditi Saha



Neurology
• Rhodemarie Maron



Dermatology
• Craig Elmets



Pulmonary
• Maria del Pilar Acosta Lara



Radiology
• Pradeep Bhambhani



Rheumatology
• Mitali Sen



Clinical Pharmacy
• Daniel Sokolowski



Nursing
• Alana Gill



Endocrinology
• Krishmita Siwakoti



Radiation Oncology
• Mehran Yusuf



Nurse Navigation
• Kellie Meloun



Patient Services Coordinator
• Melanie Heigl

Histio-Care Network



Histiocytic Disorder Follow-up Study

Participate!

If you or your loved one have ever been diagnosed with a histiocytic disorder

Earn up to \$25 for participating!

- **Goal:** To understand the various health problems experienced by patients with histiocytic disorders
- Findings from the study will lead to development of guidelines for long-term care of patients

What does the study involve?

- Answering a survey (45-60 min) about your health conditions
- Survey can be completed online or via paper form
- No need to travel- take the survey from the comfort of your home

Who can participate?

- Adults or children with ECD, LCH, RDD, or JXG
- At least 2 years since diagnosis
- Family members of those who passed away
- English-speaking

HISTIOCYTOSIS  ASSOCIATION™
A Rare Community

O'NEAL COMPREHENSIVE
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UAB MEDICINE.



If you are interested in participating or have any questions, contact our study staff:

Phone: 1-866-438-1640

Email: histio@uabmc.edu

Take away suggestions

Many types of histiocytic neoplasms (slow or fast growing)

Diagnosis of ECD is complex- relies heavily on the clinician

Important to know about LCH and RDD due to overlaps

Need to know natural history of ECD or other subtypes

Critical to participate in research (ECD Registry, Histiocytic Disorder Follow-up Study)

Acknowledgements

Patients and families

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Anita D'Souza

Histiocytosis Working Group

Research team (UAB)

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Vyshi Rallapalle
Tanvi Bhadkamkar
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Belinda Cobb

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Uplifting Athletes/ECDGA Young Investigator
Mayo Center for Individualized Medicine

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