

Erdheim-Chester Disease (ECD)

- Erdheim-Chester disease (ECD) is a rare form of non-Langerhans cell histiocytosis. It is a rare type of blood disease that belongs to the histiocytic disorders group, or histiocytosis. ECD is also classified as a hematologic neoplasm.
- Histiocytes are cells within the immune system that function as “phagocytes,” which means they ingest and remove foreign bodies like bacteria and debris. These cells, which normally help fight infection and injury, then gather in different organs and tissues and can result in a variety of symptoms, including organ failure.
- ECD was previously considered an inflammatory or autoimmune disease, but was recognized as a blood cancer in 2016 by the World Health Organization. This was due to the discovery of cancer-causing DNA changes (mutations) in BRAF and other genes in biopsy samples from most patients.
- ECD is a multisystem disease, organ systems could include ophthalmic/periorbital, pulmonary, cardiovascular, renal, musculoskeletal, dermatologic, and central nervous system, but most commonly affects the long bones of the legs around the knees.
- Due to the wide variety of manifestations of ECD often mimicking other diseases, it is believed to be under-diagnosed.
- Fewer than 2,000 diagnosed cases are estimated worldwide.
- Typical onset is between 40 and 70 years of age, although there are documented cases in all age groups; with a slight preponderance in males.
- More than 50% of patients have the BRAF V600E mutation and the majority of the remaining patients have other mutations in the MAPK signaling pathway.
- Chronic uncontrolled inflammation is an important mediator of disease pathogenesis.
- While the exact cause of developing ECD is unknown, it is not considered a hereditary or contagious disease.
- Diagnosis requires a biopsy, scans, and clinical symptom assessment.

**Learn more about
ECD and help us
spread awareness!**

