

Overview of ECD Basic Biology

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ECD Clinical presentations

Neuro-endocrine 37%

Polyuria, polydipsia 25%,
Ataxia 46%, dysmetria 33%
Cranial neuropathy 56%,
Peripheral neuropathy 56%
Pyramidal tract signs 30%
Seizures 8%
Cognitive Dysfunction 52%

Ophthalmic

Oculomotor abnormalities 44%
Exophthalmos 27%
Xanthelasma 22%

Cardiovascular

Cardiac Failure 17%

Genitourinary

Renal failure 10%
Hydronephrosis 27%
Hypogonadism 60%

Hematological

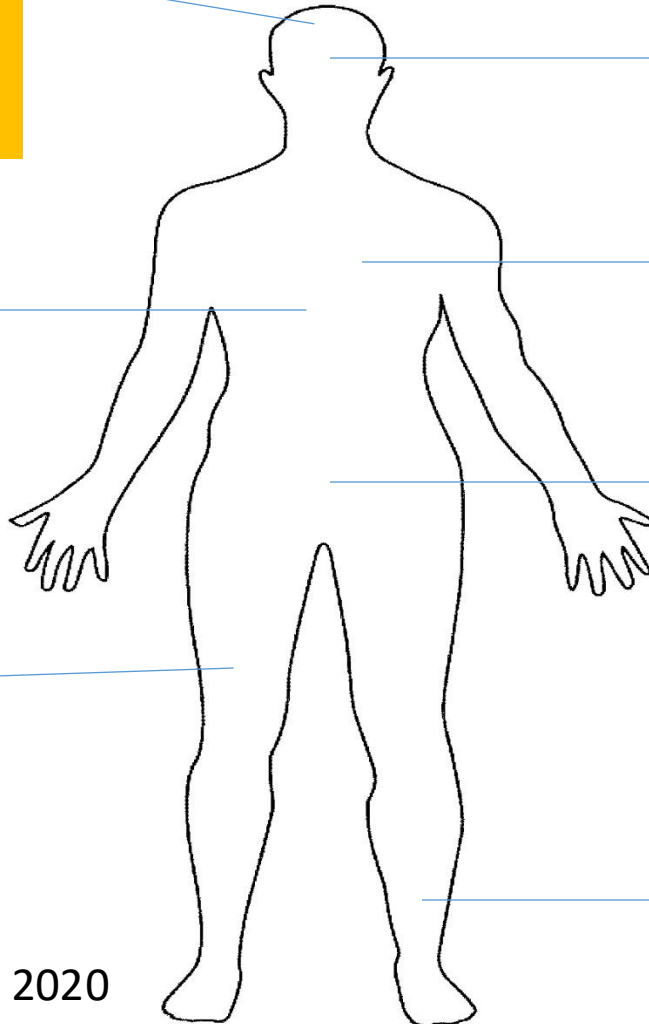
Clonal hematopoiesis 43%
Hematologic malignancy 15%

Pulmonary

Dyspnea 30%

Musculoskeletal

Bone pain 38%



Haroche Blood 2020; Chen Aubart Blood 2020

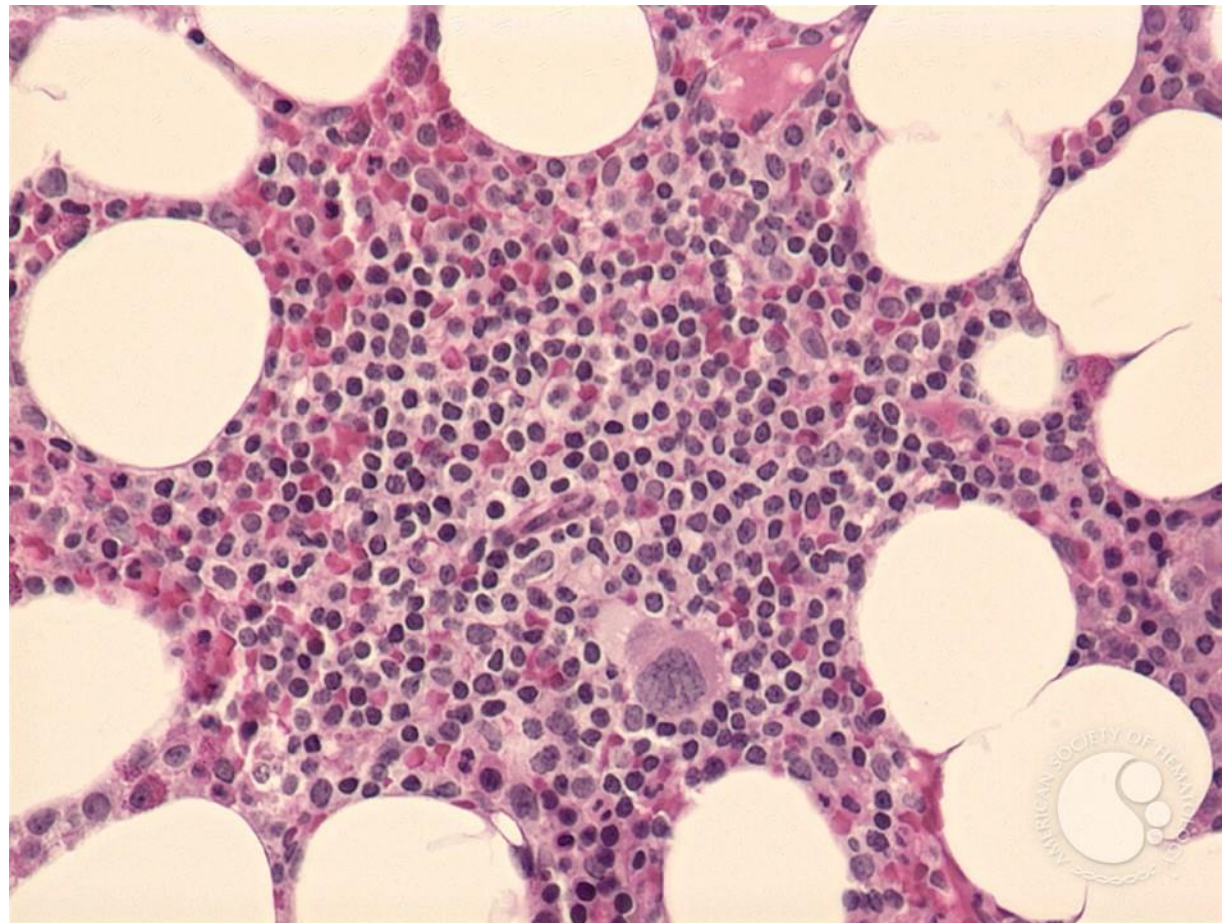
Louisa Boyd Ann Neurolo 2020, Ghotra AS, Echocardiography 2019;36:229

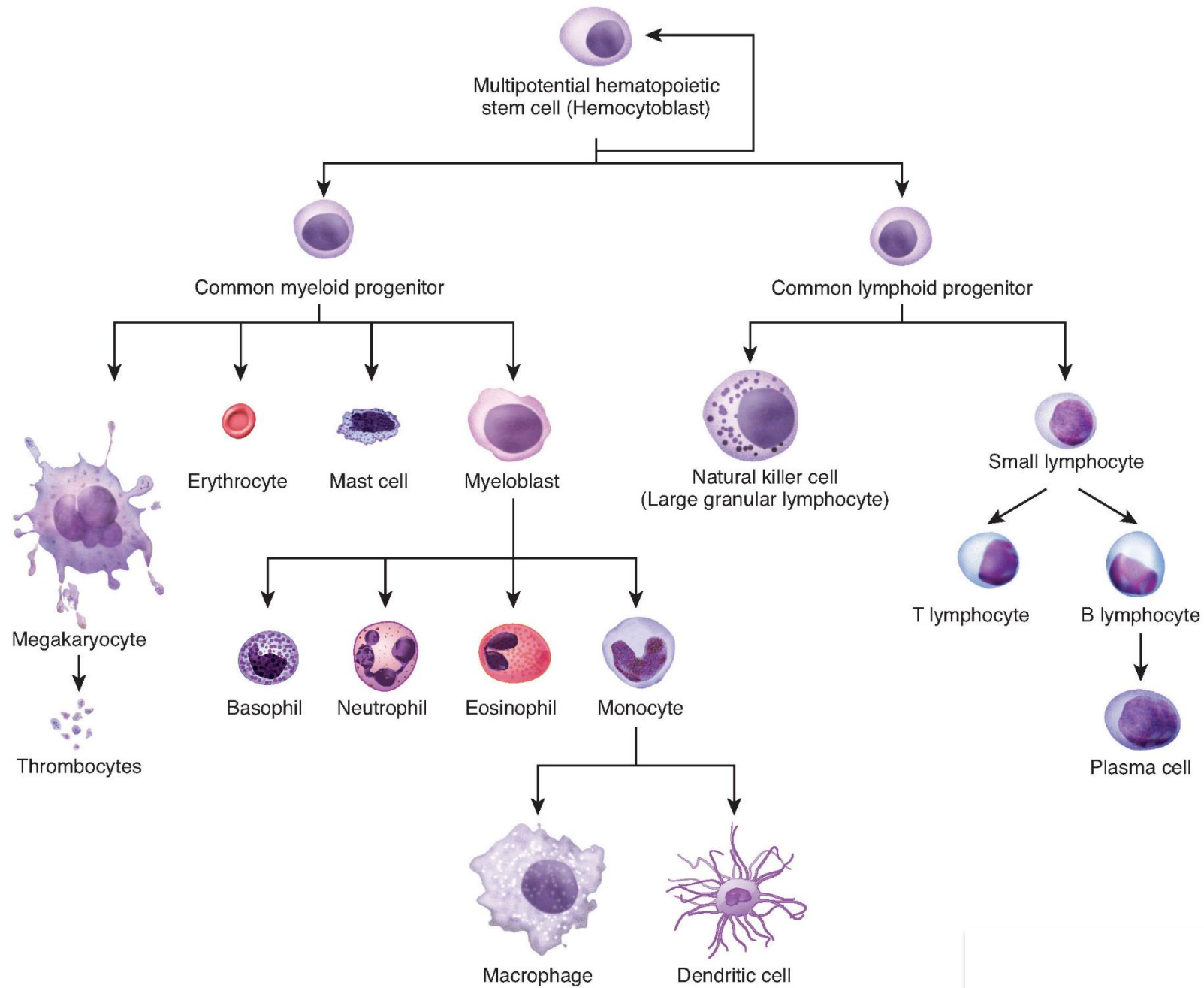
Normal adult bone marrow – trephine biopsy - 2.

Image ID: 4243

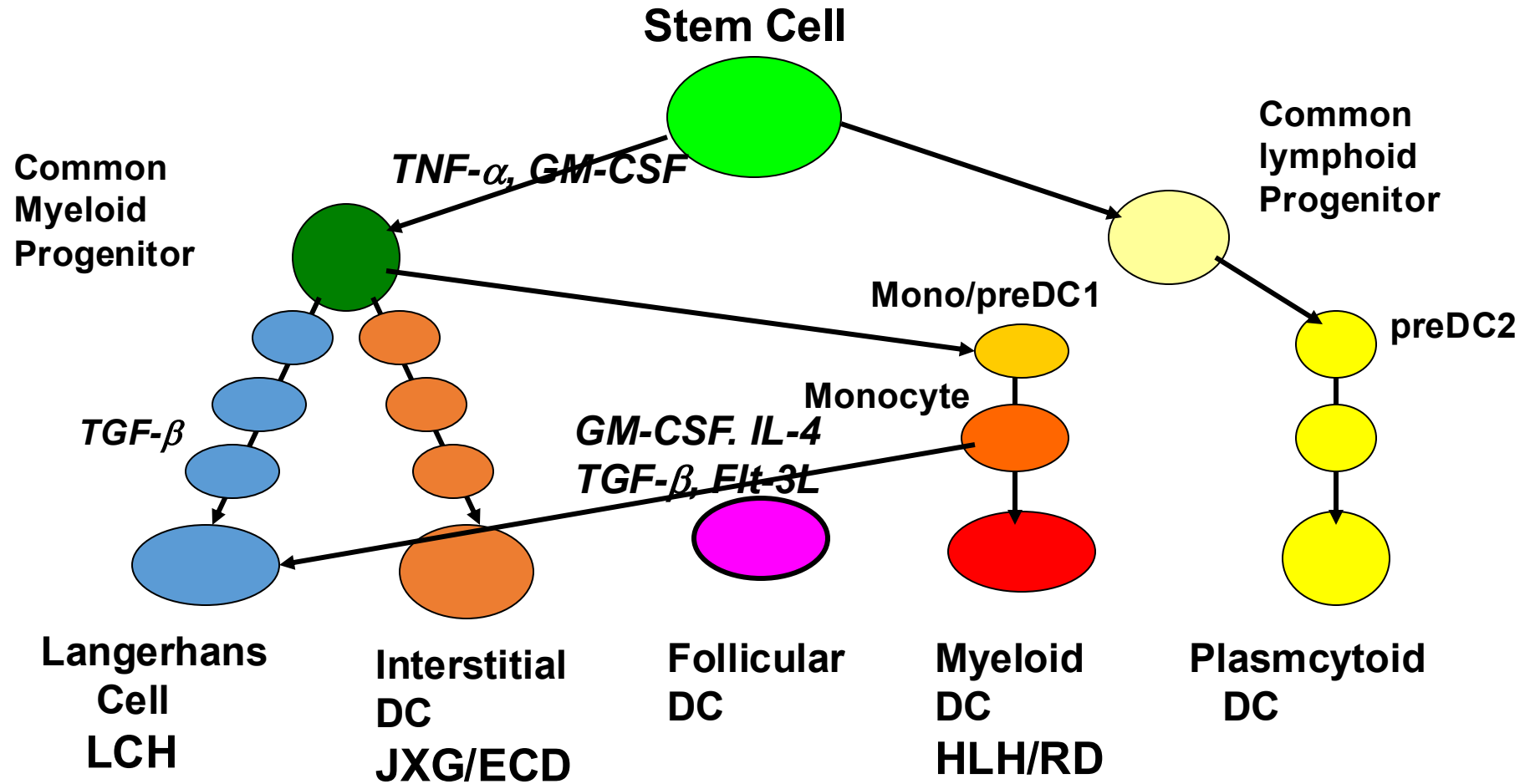
Authors: Peter Maslak

Category: Laboratory Hematology > Normal and reactive bone marrow

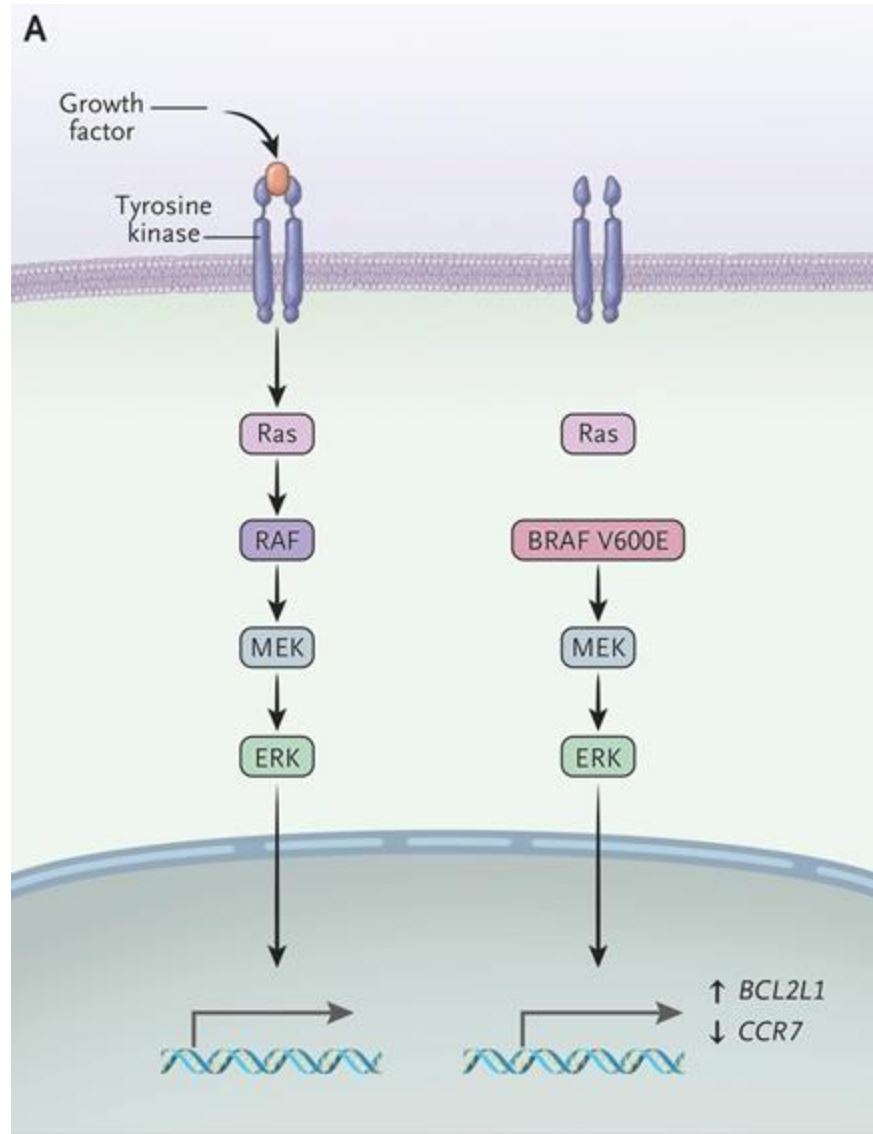




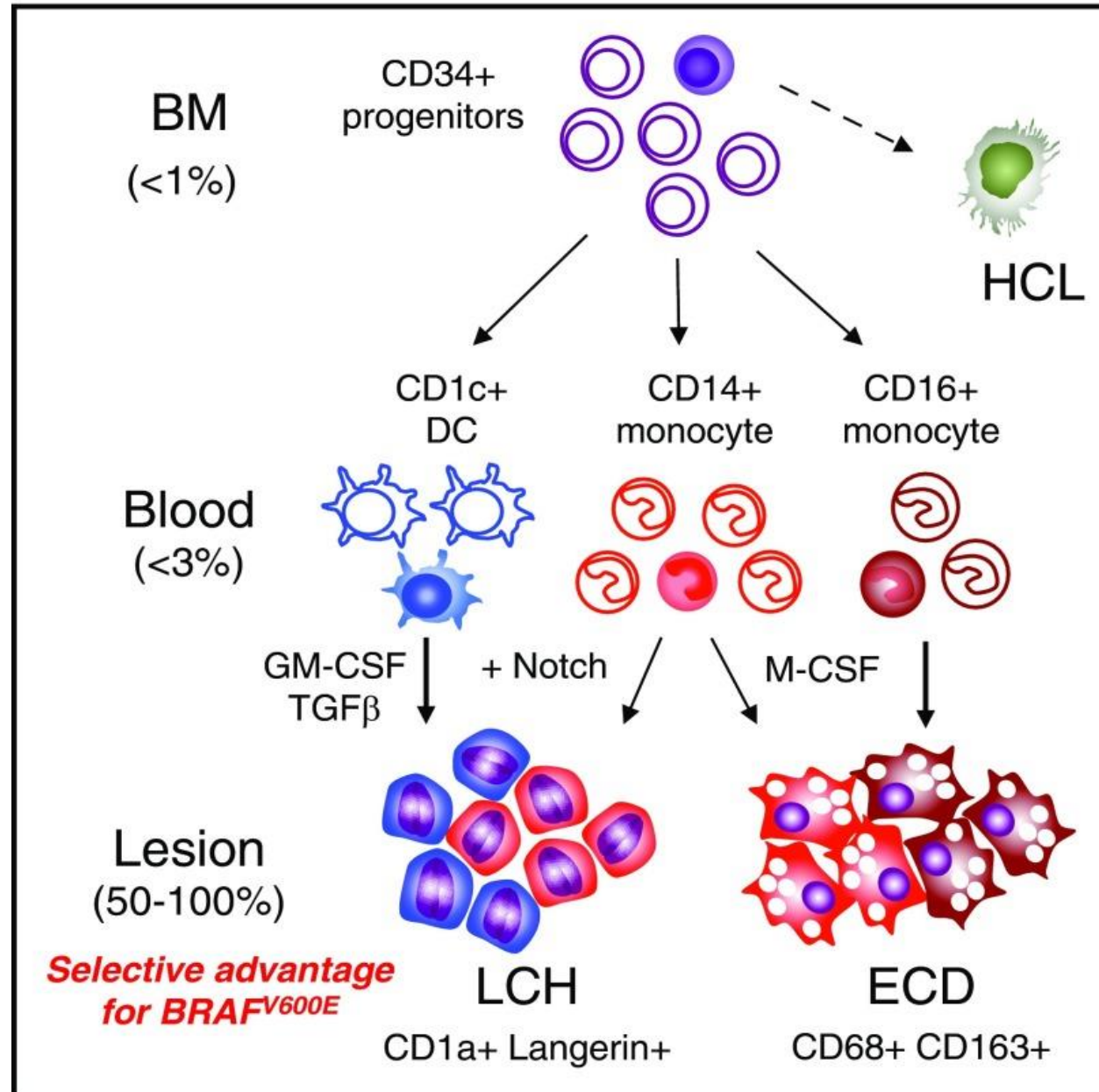
Myeloid Cell Development, Circa 2005



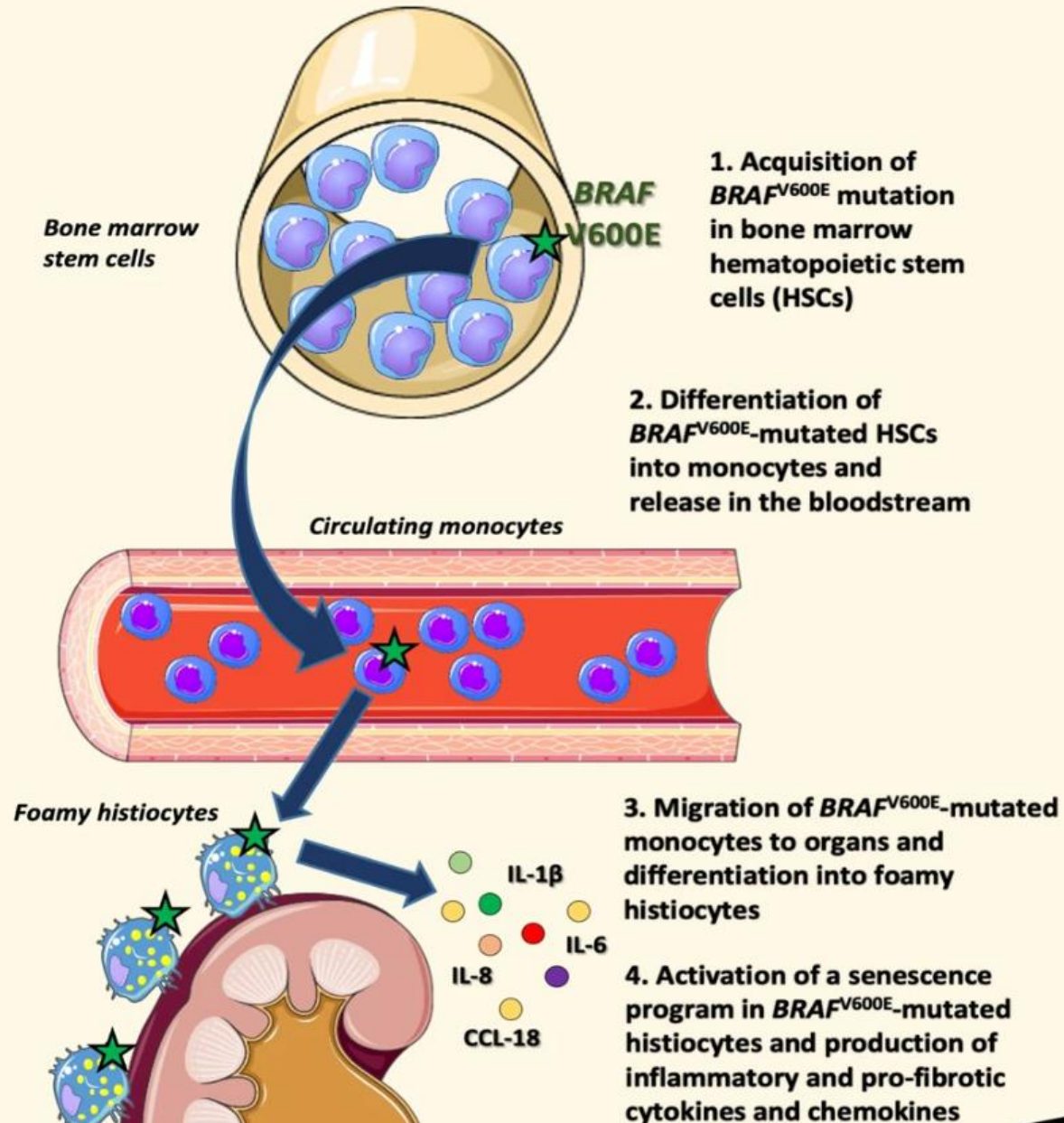
What is the BRAFV600E Mutation and How Does It Affect Cells?



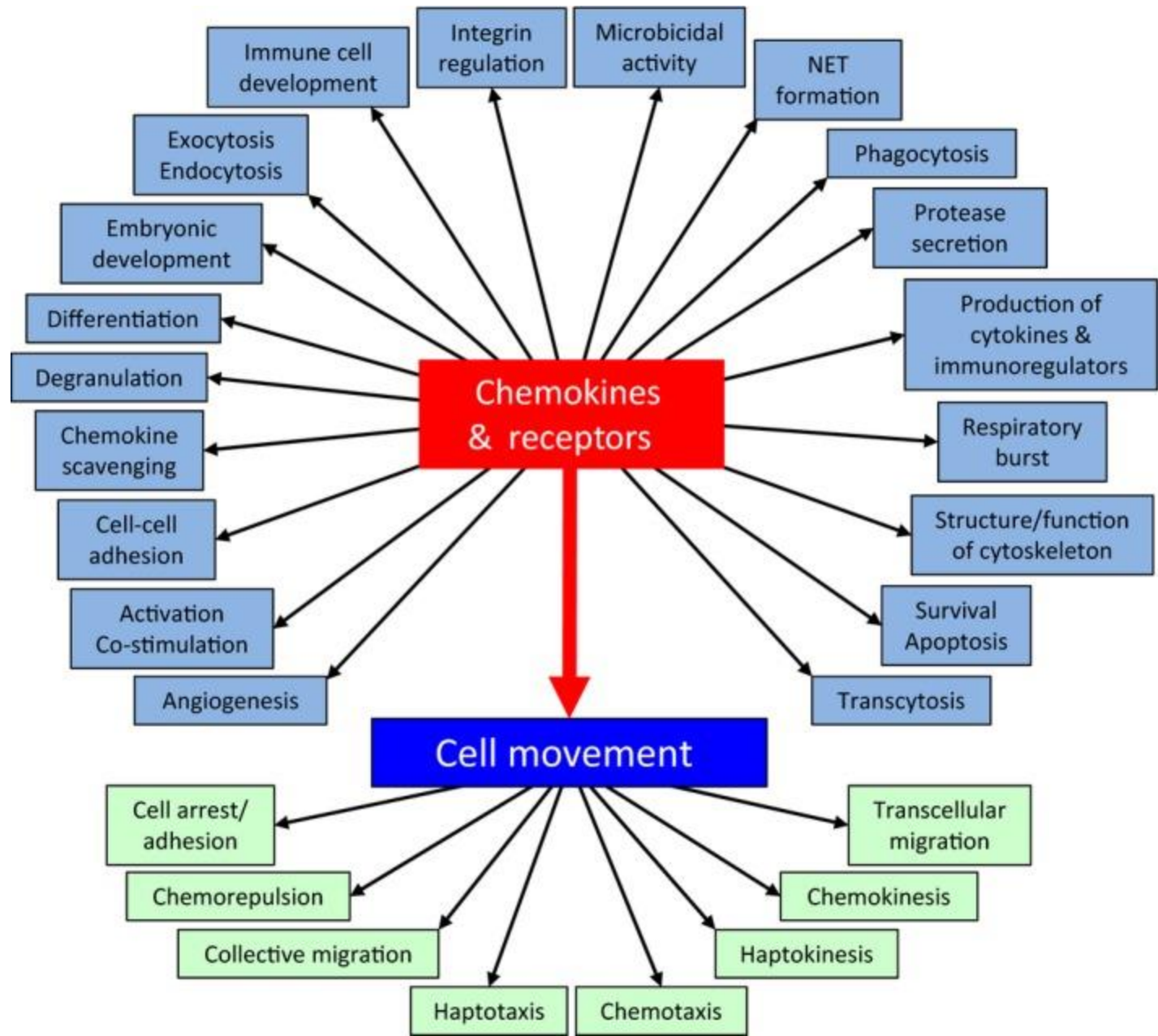
Origin of ECD- 2017



Ontogenesis of mutated histiocytes in Erdheim-Chester Disease



Used with permission
Pegoraro F, et al
Leukemia 2020



Add Cytokines ECD Signs & Symptoms

Chemical messengers that tell cells how to behave

Cell activation: Cytokines tell cells where to go and what to do. For example, cytokines can direct immune cells toward an infection site so the cells can fight germs there. They can heighten the processes associated with inflammation.

Cell differentiation: Cytokines can tell immature cells to develop into a specific type of cell. For example, cytokines can tell an immature cell to mature into a white blood cell capable of fighting infection.

Cell proliferation: Cytokines can tell a cell to make more cells just like it. For example, cytokines can tell a white blood cell to make more white blood cells to fight infection: make inflammatory cytokines

ECD: a biologic stew that drives supposedly normal immune cells to create the diverse pathologic effects patients suffer from