

Gene name: ELANE

External Ids for ELANE Gene: HGNC: [3309](#) NCBI Gene: [1991](#) Ensembl: [ENSG00000197561](#) OMIM®: [130130](#) UniProtKB/Swiss-Prot: [P08246](#)

NCBI Gene Summary for ELANE Gene: Elastases form a subfamily of serine proteases that hydrolyze many proteins in addition to elastin. Humans have six elastase genes which encode structurally similar proteins. The encoded preproprotein is proteolytically processed to generate the active protease. Following activation, this protease hydrolyzes proteins within specialized neutrophil lysosomes, called azurophil granules, as well as proteins of the extracellular matrix. The enzyme may play a role in degenerative and inflammatory diseases through proteolysis of collagen-IV and elastin. This protein also degrades the outer membrane protein A (OmpA) of E. coli as well as the virulence factors of such bacteria as Shigella, Salmonella and Yersinia. Mutations in this gene are associated with cyclic neutropenia and severe congenital neutropenia (SCN). This gene is present in a gene cluster on chromosome 19.

GeneCards Summary for ELANE Gene: ELANE (Elastase, Neutrophil Expressed) is a Protein Coding gene. Diseases associated with ELANE include [Cyclic Neutropenia](#) and [Neutropenia, Severe Congenital, 1, Autosomal Dominant](#). Among its related pathways are [Complement cascade](#) and [Innate Immune System](#). Gene Ontology (GO) annotations related to this gene include *serine-type endopeptidase activity* and *peptidase activity*. An important paralog of this gene is [PRTN3](#).

UniProtKB/Swiss-Prot Summary for ELANE Gene: Serine protease that modifies the functions of natural killer cells, monocytes and granulocytes. Inhibits C5a-dependent neutrophil enzyme release and chemotaxis (PubMed:[15140022](#)). Promotes cleavage of GSDMB, thereby inhibiting pyroptosis (PubMed:[36899106](#)).

The **ELANE** gene encodes neutrophil elastase, a serine protease predominantly expressed in neutrophils. This enzyme plays a crucial role in the innate immune response by degrading pathogens and modulating inflammatory processes.

Function in Sepsis: In sepsis, an excessive and dysregulated immune response to infection, neutrophil elastase contributes to both pathogen clearance and tissue damage. Elevated activity of neutrophil elastase can lead to the degradation of extracellular matrix components and endothelial damage, exacerbating inflammation and contributing to organ dysfunction.

Pathways Involved in Sepsis:

Neutrophil elastase is involved in several key pathways during sepsis:

- **Neutrophil Extracellular Trap (NET) Formation:** Neutrophil elastase is essential for the formation of NETs, which trap and neutralize pathogens. However, excessive NET formation can contribute to tissue injury and has been implicated in the pathogenesis of sepsis.
- **Pyroptosis:** Recent studies have identified ELANE as part of a pyroptosis-related gene signature associated with sepsis prognosis, indicating its involvement in inflammatory cell death pathways.

Diagnostic and Prognostic Role:

- **Diagnostic Role:** While ELANE expression is elevated in sepsis, its specificity as a standalone diagnostic biomarker is limited due to its involvement in various inflammatory conditions.

However, it may contribute to multi-gene diagnostic panels aimed at improving sepsis detection.

- **Prognostic Role:** Elevated ELANE expression has been associated with poorer outcomes in sepsis patients. Studies have developed prognostic risk scores incorporating ELANE and other genes to stratify sepsis patients based on survival probabilities.

In summary, the ELANE gene, through its product neutrophil elastase, plays a significant role in the immune response during sepsis. Its involvement in key inflammatory pathways and association with patient outcomes highlight its potential as a component of diagnostic and prognostic tools in sepsis management.

Neutrophil trap formation:

