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C16orf57 Antibody Blocking Peptide

Catalog Number: bs-9631P
Activity: Not tested
Purification: HPLC
Storage: Shipped at 4°C. Stored at -20°C for one year. Avoid repeated freeze/thaw cycles.
Background: Involvement in disease;Defects in C16orf57 are the cause of poikiloderma with neutropenia (PN). PN is a genodermatosis characterized by poikiloderma, pachyonychia and chronic neutropenia. The disorder starts as a papular erythematous rash on the limbs during the first year of life. It gradually spreads centripetally and, as the papular rash resolves, hypo- and hyperpigmentation result, with development of telangiectasias. Another skin manifestation is pachyonychia, but alopecia and leukoplakia are distinctively absent. One of the most important extracutaneous symptoms is an increased susceptibility to infections, mainly affecting the respiratory system, primarily due to a chronic neutropenia and to neutrophil functional defects. Bone marrow abnormalities account for neutropenia and may evolve into myelodysplasia associated with the risk of leukemic transformation. Poikiloderma with neutropenia shows phenotypic overlap with Rothmund-Thomson syndrome.