Cochlin is a secreted protein encoded by the coagulation factor C homology (COCH) gene, a cochlear gene. It constitutes 70% of the inner ear proteins and is classified into three glycosylated isoforms: p63s, p44s and p40. Cochlin contains an N-terminal LCCL domain and two von Willebrand factor A-like domains. Mutations in the COCH gene cause DFNA9, an autosomal dominant nonsyndromic auditory and vestibular dysfunction disorder, as a result of either an amino acid deletion in the LCCL domain or missense substitutions. Microfibrillar deposits accumulate in the inner ear of individuals with DFNA9 and these deposits may contain the Cochlin protein. Cochlin is a target antigen for autoimmune sensorineural hearing loss.

REFERENCES

CHROMOSOMAL LOCATION
Genetic locus: COCH [human] mapping to 14q12-q13; Coch (mouse) mapping to 12 C1.

SOURCE
Cochlin (H-80) is a rabbit polyclonal antibody raised against amino acids 401-480 mapping near the C-terminus of Cochlin of human origin.