

Product datasheet

Human CLC7 peptide ab32492

Overview

Product name Human CLC7 peptide

Description

Nature Synthetic

Amino Acid Sequence

Species Human

Specifications

Our [Abpromise guarantee](#) covers the use of **ab32492** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Form Liquid

Additional notes

- First try to dissolve a small amount of peptide in either water or buffer. The more charged residues on a peptide, the more soluble it is in aqueous solutions.
- If the peptide doesn't dissolve try an organic solvent e.g. DMSO, then dilute using water or buffer.
- Consider that any solvent used must be compatible with your assay. If a peptide does not dissolve and you need to recover it, lyophilise to remove the solvent.
- Gentle warming and sonication can effectively aid peptide solubilisation. If the solution is cloudy or has gelled the peptide may be in suspension rather than solubilised.
- Peptides containing cysteine are easily oxidised, so should be prepared in solution just prior to use.

Preparation and Storage

Stability and Storage Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles.

Information available upon request.

General Info

Function	Mediates the exchange of chloride ions against protons. Functions as antiporter and contributes to the acidification of the lysosome lumen.
Tissue specificity	Brain, testis, muscle and kidney.
Involvement in disease	<p>Defects in CLCN7 are the cause of osteopetrosis autosomal recessive type 4 (OPTB4) [MIM:611490]; also known as infantile malignant osteopetrosis type 2. Osteopetrosis is a rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood.</p> <p>Defects in CLCN7 are the cause of osteopetrosis autosomal dominant type 2 (OPTA2) [MIM:166600]; also known as autosomal dominant Albers-Schonberg disease or marble disease autosomal dominant. Osteopetrosis is a rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. The disorder occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTA2 is the most common form of osteopetrosis, occurring in adolescence or adulthood. It is characterized by sclerosis, predominantly involving the spine, the pelvis and the skull base.</p>
Sequence similarities	Belongs to the chloride channel (TC 2.A.49) family. CIC-7/CLCN7 subfamily. Contains 2 CBS domains.
Cellular localization	Lysosome membrane.

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