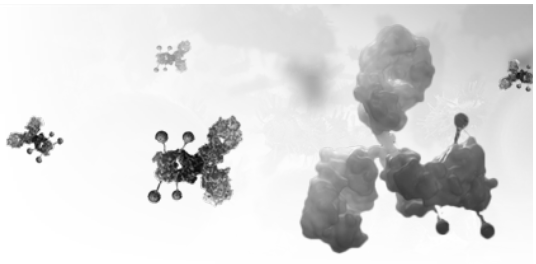

GJB6 peptide (DAG-P0555)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

Antigen Description	Defects in GJB6 are the cause of ectodermal dysplasia type 2 (ED2); also known as Clouston syndrome. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ED2 is an autosomal dominant condition characterized by atrichosis, nail hypoplasia and deformities, hyperpigmentation of the skin, normal teeth, normal sweat and sebaceous gland function. Palmoplantar hyperkeratosis is a frequent features. Hearing impairment has been detected in few cases of ED2. Defects in GJB6 are the cause of deafness autosomal recessive type 1B (DFNB1B) [MIM:612645]. DFNB1B is a form of sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. Defects in GJB6 are the cause of deafness autosomal dominant type 3B (DFNA3B).
Purity	70 - 90% by HPLC.
Conjugate	Unconjugated
Sequence Similarities	Belongs to the connexin family. Beta-type (group I) subfamily.
Preservative	None
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.

GENE INFORMATION

Gene Name	Gjb6 gap junction protein, beta 6 [Mus musculus (house mouse)]
Official Symbol	GJB6
Synonyms	GJB6; gap junction protein, beta 6; Cx30; AA958971; D14Bwg0506e; gap junction beta-6 protein; connexin-30; gap junction membrane channel protein beta 6;

45-1 Ramsey Road, Shirley, NY 11967, USA
Email: info@creative-diagnostics.com
Tel: 1-631-624-4882 Fax: 1-631-938-8221

Entrez Gene ID	14623
mRNA Refseq	NM_001010937.2
Protein Refseq	NP_001010937.1
UniProt ID	P70689
Chromosome Location	14 C3; 14 30.1 cM
Pathway	Calcium Regulation in the Cardiac Cell, organism-specific biosystem; XPodNet - protein-protein interactions in the podocyte expanded by STRING, organism-specific biosystem;
Function	protein binding;

45-1 Ramsey Road, Shirley, NY 11967, USA
Email: info@creative-diagnostics.com
Tel: 1-631-624-4882 Fax: 1-631-938-8221