

## DESCRIPTION

<b>Source</b>	Mouse myeloma cell line, NS0-derived		
	Human Ectodermal Dysplasia Receptor (Glu27 - Ile189) Accession # Q9UNE0	DIEGRMD	Human IgG <sub>1</sub> (Pro100 - Lys330)
	N-terminus		C-terminus

**N-terminal Sequence Analysis** Glu27

**Structure / Form** Disulfide-linked homodimer

**Predicted Molecular Mass** 44.3 kDa (monomer)

## SPECIFICATIONS

**SDS-PAGE** 55-60 kDa, reducing conditions

**Activity** Measured by the ability of the immobilized rhEDAR/Fc Chimera to bind biotinylated rhEDA-A1/His in a functional ELISA.  
**Optimal dilutions should be determined by each laboratory for each application.**

**Endotoxin Level** <1.0 EU per 1 µg of the protein by the LAL method.

**Purity** >90%, by SDS-PAGE under reducing conditions and visualized by silver stain.

**Formulation** Lyophilized from a 0.2 µm filtered solution in PBS. See Certificate of Analysis for details.

## PREPARATION AND STORAGE

**Reconstitution** Reconstitute at 100 µg/mL in sterile PBS.

**Shipping** The product is shipped at ambient temperature. Upon receipt, store it immediately at the temperature recommended below.

**Stability & Storage** Use a manual defrost freezer and avoid repeated freeze-thaw cycles.

- 12 months from date of receipt, -20 to -70 °C as supplied.
- 1 month, 2 to 8 °C under sterile conditions after reconstitution.
- 3 months, -20 to -70 °C under sterile conditions after reconstitution.

## BACKGROUND

EDAR is a type I transmembrane protein which is a member of the TNF Receptor Superfamily (TNFRSF). The extracellular domain contains 14 cysteine residues, six of which approximate the TNFRSF cysteine-rich region; the cytoplasmic domain contains a region with homology to the death domains found in other TNFRSF members. Human EDAR is a 488 amino acid (aa) protein with a predicted 26 aa signal, a 163 aa extracellular domain, a 22 aa transmembrane domain, and a 277 aa cytoplasmic domain. The human and mouse EDAR homologs share 91% identity. Within the TNFRSF, EDAR shares the highest homologies with XEDAR and TNFRSF19/TROY. EDA-A1 is the EDAR ligand. EDA and EDAR have been associated with hypohidrotic ectodermal dysplasia (HED). HED is characterized by abnormalities in hair, teeth and eccrine sweat gland morphogenesis. HED was initially found to associate with two gene loci, *tabby* and *downless*. *Tabby* was later identified as the gene for EDA and *downless* as the autosomal EDAR gene. EDA has two splice variants, EDA-A1 and EDA-A2, which differ by only two amino acids. Despite this minor difference, the EDA isoforms display strong receptor specificity. EDA-A1 only binds EDAR, whereas EDA-A2 binds to XEDAR, an X-linked TNFRSF member with high homology to EDAR. Mutations in EDA, EDAR and XEDAR have been associated with HED.

### References:

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3. Monreal, A.W. *et al.* (1999) Nat. Genet. **22**:366.
4. Schneider, P. *et al.* (2001) J. Biol. Chem. **276**:18819.
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6. Yan, M. *et al.* (2000) Science **290**:523.