

2282del4 MUTATION IN THE FILAGGRIN GENE

ORDERING INFORMATION

REF: GEN-052-25
Tests: 25 Reactions: 31
Manufacturer: BioMol Laboratories s.r.l.

CONTENTS OF THE KIT

The kit consists of reagents for
Real-Time PCR amplification
*reagents for the extraction of
genomic DNA are not supplied in
the kit



PRODUCT CHARACTERISTICS

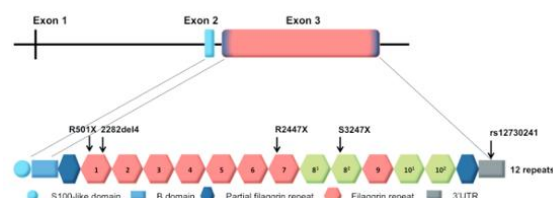
Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS-RESEARCH USE ONLY**. Determination of genetic mutation 2282del4 of the filaggrin gene (rs558269137) by Real-Time PCR technique. Kit optimized for Real-time PCR instruments Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.

SCIENTIFIC BACKGROUND

Atopic dermatitis (AD) is a chronic, recurrent inflammatory skin disease that is commonly widespread and usually develops in infancy or early childhood, characterised by dry skin, eczema-like dermatitis and persistent itching. Itching leads to reduced sleep quality, seriously affecting the health of the child and their family. The mechanisms underlying the pathogenesis of AD are complex; much evidence attributes it to the interaction of genetic and environmental factors, resulting in altered skin barrier function, immune system disorders and epidermal microflora imbalance. Filaggrin is one of the most important proteins involved in epidermal barrier homeostasis. The filaggrin (FLG) gene is a complex, highly polymorphic and repetitive gene located in the epidermal differentiation complex on chromosome 1q21. Null mutations in FLG have been associated with earlier onset of the disease.

CLINICAL SIGNIFICANCE

Filaggrin is one of the main structural proteins in the stratum corneum (SC) and is generated from the precursor profilagin, which is expressed in keratinocytes undergoing terminal differentiation and is the main constituent of keratohyalin granules in the granular layer. Forty-nine truncating mutations, including the 2282del4 (rs558269137) mutation, have been described along the entire length of the profilagin molecule, and the incidence of these mutations varies considerably between different ethnic groups and populations. This mutation is the best known among a relatively large number of loss-of-function (LoF) mutations in the FLG gene that increase the risk of ichthyosis vulgaris, atopic dermatitis and eczema.



§ Association between polymorphisms and atopic dermatitis susceptibility: A systematic review and meta-analysis. Gene 913 (2024) 148397 <https://doi.org/10.1016/j.gene.2024.148397>

§ The Value of FLG Null Mutations in Predicting Treatment Response in Atopic Dermatitis: An Observational Study in Finnish Patients. Acta Derm Venereol 2017; 97: 456–463. doi:10.2340/00015555-2578.

§ The Genetics and Epigenetics of Atopic Dermatitis—Filaggrin and Other Polymorphisms. Clinic Rev Allerg Immunol (2016) 51:315–328 DOI 10.1007/s12016-015-8508-5

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DESCRIPTION	LABEL	VOLUME	STORAGE
GEN-052-25			
Mix oligonucleotides and probes	Mix FLG 10X	1 x 62 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 310 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA	Control 1	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 2	1 x 22 µl	-20°C
Genomic DNA or recombinant DNA	Control 3	1 x 22 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-052-25

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissue, cells
POSITIVE CONTROLS	Recombinant DNA for at least 3 analytical sessions
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx e Agilent AriaDx, Thermofisher QuantStudio™ 5 Real-Time PCR System.
TECHNOLOGY	Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels
RUNNING TIME	85 min
THERMAL CYCLING PROFILE	1 cycle at 95 °C (10 min); 45 cycles at 95 °C (15 sec) + 60 °C (60 sec)
ANALYTICAL SPECIFICITY	Absence of non-specific pairings oligonucleotides and probes; absence of cross-reactivity
LIMIT OF DETECTION (LOD)	≥ 5 ng of genomic DNA
LIMIT OF BLANK (LOB)	>40 Cq
REPRODUCIBILITY	99,9%
SPECIFICITY / SENSITIVITY	99,9%/98%