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## Substantiating atypical phenotypes of epidermolysis bullosa

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# **SUBSTANTIATING ATYPICAL PHENOTYPES OF EPIDERMOLYSIS BULLOSA**

## **Proefschrift**

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## LIST OF ABBREVIATIONS

ABD	actin binding domain
AI	amelogenesis imperfecta
AD	autosomal dominant
AF	anchoring fibrils
APSS	acral peeling skin syndrome
AR	autosomal recessive
BMZ	basement membrane zone
cDNA	copy DNA
DDEB	dominant dystrophic epidermolysis bullosa
DEB	dystrophic epidermolysis bullosa
EB	epidermolysis bullosa
EBS	epidermolysis bullosa simplex
EBS-AR	exophilin 5: epidermolysis bullosa autosomal recessive exophilin 5
EBS-gen sev	epidermolysis bullosa simplex generalized severe
EBS-MD	epidermolysis bullosa simplex with muscular dystrophy
EBS-MD-Mys	epidermolysis bullosa simplex with muscular dystrophy and myasthenic syndrome
EBP	epidermolysis bullosa pruriginosa
EBP-excoriée	epidermolysis bullosa pruriginosa excoriee
EBS-Og	epidermolysis bullosa simplex Ogna
EBS-PA	epidermolysis bullosa simplex with pyloric atresia
EBS-plectin1a	epidermolysis bullosa simplex plectin isoform 1 <sup>a</sup> deficiency
GABEB	generalized atrophic benign epidermolysis bullosa
HD	hemidesmosome
IF	immunofluorescence antigen mapping
JEB	junctional epidermolysis bullosa
JEB-gen-intermed	junctional epidermolysis bullosa generalized intermediate (former JEB-non Hertz) type
JEB-gen sev	junctional epidermolysis bullosa generalized severe (earlier JEB-Hertz) type
JEB-I	junctional epidermolysis bullosa inversa
JEB-LO	junctional epidermolysis bullosa late onset
JEB-loc	junctional epidermolysis bullosa localized
JEB-PA	junctional epidermolysis bullosa with pyloric atresia
JEB-RR	junctional epidermolysis bullosa respiratory and renal
KS	kindler syndrome
Laminin 332	LM-332
LGMD2Q	limb girdle muscular dystrophy
MOI	mode of inheritance
mRNA	messenger RNA
NC1	non collagenous 1 domain
NC2	non collagenous 2 domain
NGS	next generation sequencing



PCR	polymerase chain reaction
P1a	plectin isoform 1a
P1c	plectin isoform 1c
P1f	plectin isoform 1f
RDEB	recessive dystrophic epidermolysis bullosa
RDEB-gen sev	recessive dystrophic epidermolysis bullosa generalized severe type
SS	Sanger sequencing
TEM	transmission electron microscopy
WES	Whole exome sequencing
WGS	Whole genome sequencing



