

# CONTENTS

---

<b>1</b>	<b>GENERAL ASPECTS</b>	
1.1	Definition – <i>Christoph M. Lanschuetzer</i> . . . . .	3
1.2	Classification and molecular basis of hereditary epidermolysis bullosa – <i>Christoph M. Lanschuetzer, Jo-David Fine</i> . . . . .	6
1.3	Epidemiology of inherited epidermolysis bullosa – <i>Jo-David Fine</i> . . . . .	24
1.4	Diagnosis	
1.4.1	Non-molecular techniques	
1.4.1.1	Routine histopathology in epidermolysis bullosa – <i>Martin Laimer</i> . . . . .	30
1.4.1.2	Antigen mapping – <i>Gabriela Pohla-Gubo, Elke Nischler, Helmut Hintner</i> . . . . .	35
1.4.1.3	Electron microscopy for the diagnosis of epidermolysis bullosa – <i>Robin A. J. Eady</i> . . . . .	43
1.4.2	Molecular techniques	
1.4.2.1	Mutation analysis – <i>Alfred Klausegger, Johann Bauer</i> . . . . .	54
1.4.2.2	Mosaicism in epidermolysis bullosa – <i>Martin Laimer</i> . . . . .	65
1.4.2.3	Prenatal and preimplantation genetic diagnosis in epidermolysis bullosa – <i>Hiva Fassihi, John McGrath</i> . . . . .	77
1.4.3	Genetic counseling – <i>Johann Bauer</i> . . . . .	89
<b>2</b>	<b>CLINICAL MANIFESTATIONS AND COMPLICATIONS</b>	
2.1	Cutaneous	
2.1.1	General cutaneous manifestations – <i>Jo-David Fine</i> . . . . .	99
2.1.2	Epidermolysis bullosa naevi – <i>Christoph M. Lanschuetzer</i> . . . . .	107
2.1.3	Epidermolysis bullosa and cancer – <i>Jo-David Fine, Christoph M. Lanschuetzer</i> . . . . .	116

2.2	Extracutaneous	
2.2.1	Ophthalmologic aspects of epidermolysis bullosa – <i>Josef Stoiber</i> . . . . .	132
2.2.2	Ear, nose, and throat complications – <i>Martin Laimer</i> . . .	143
2.2.3	Intraoral disease – <i>Martin Laimer, Elke Nischler</i> . . . . .	150
2.2.4	Gastrointestinal complications – <i>Elke Nischler</i> . . . . .	167
2.2.5	Musculoskeletal deformities – <i>Jo-David Fine</i> . . . . .	177
2.2.6	Other internal complications – <i>Jo-David Fine</i> . . . . .	185
2.2.7	Premature death in epidermolysis bullosa – <i>Jo-David Fine</i> . . . . .	197
2.2.8	Psychological and sociological aspects – <i>Jo-David Fine</i> . . . . .	204
<b>3</b>	<b>THERAPEUTICAL APPROACHES</b>	
3.1	Dermatologic and medical	
3.1.1	Wound healing – <i>Christoph M. Lanschuetzer</i> . . . . .	211
3.1.2	Pain management in epidermolysis bullosa – <i>Elke Nischler</i> . . . . .	227
3.1.3	Pruritus in epidermolysis bullosa – <i>Elke Nischler, Anja Diem</i> . . . . .	233
3.2	debra-austria and the eb-haus Austria – <i>Gabriela Pohla-Gubo, Rainer Riedl, Helmut Hintner</i> . . . . .	241
3.3	Surgical interventions – <i>Barbara Ludwikowski</i> . . . . .	246
3.4	Nutritional support for children with epidermolysis bullosa – <i>Lesley Haynes</i> . . . . .	258
3.5	Physical medicine and epidermolysis bullosa – <i>Margret Burger-Rafael</i> . . . . .	278
3.6	Molecular therapy of epidermolysis bullosa – <i>Martin Laimer, Johann W. Bauer</i> . . . . .	287
<b>4</b>	<b>LIVING WITH EB</b>	
4.1	Surveillance for extracutaneous complications – <i>Jo-David Fine</i> . . . . .	311
4.2	Living with EB – impact on daily life – <i>Anja Diem</i> . . . . .	313
	<b>INDEX</b> . . . . .	335