

# Contents

|                                                                                                     |     |
|-----------------------------------------------------------------------------------------------------|-----|
| Preface                                                                                             | vii |
| Abbreviations and glossary                                                                          | ix  |
| 1 Haemoglobin and the genetics of haemoglobin synthesis                                             | 1   |
| 2 Laboratory techniques for the identification of abnormalities of globin chain synthesis           | 33  |
| 3 $\alpha$ , $\beta$ , $\delta$ and $\gamma$ thalassaemias and related conditions                   | 93  |
| 4 Sick cell haemoglobin and its interactions with other variant haemoglobins and with thalassaemias | 209 |
| 5 Other significant haemoglobinopathies                                                             | 299 |
| 6 Acquired abnormalities of globin chain synthesis or haemoglobin structure                         | 377 |
| 7 Organisation of a haemoglobinopathy diagnostic service                                            | 403 |
| 8 Self-assessment – test cases                                                                      | 423 |
| Appendix: electronic resources                                                                      | 467 |
| Index                                                                                               | 471 |

