Handbook of FAQs on

Basic Genetics and Common Genetic Disorders for Students

This handy book aims to serve both undergraduate and postgraduate medical students in providing the essential knowledge of genetics and guidance in focused, pointed and systematic way for preparing the question-answers related to the genetics which are frequently asked in the examinations.

Key Features

- Comprehensive, illustrative, latest coverage of all the topics related to basic genetics, common genetic disorders, from the recommended academic syllabus, questions from previous question bank with suitable examples in an easy flowing question-answer format.
- Complex terminology and hard-to-grasp concepts of genetics are presented in student-friendly, simplified and understandable language.
- Up-to-date content offers a glimpse into the latest diagnostic and therapeutic advances in genetics.
- Recent genetics websites, clinical genetic databases and tables of various genetic syndromes and other disorders are provided in the appendix.
- Contributions from leading experts in the field of genetics.

Chief Editor

Komal Uppal DCH, DNB (Pediatrics), FSIAMG, PGCCMGC is currently Medical Scientist, Division of Genetics, Department of Pediatrics, All India Institute of Medical Sciences, New Delhi. She graduated from Government Medical College, Amritsar, and obtained her DCH from Dayanand Medical College, Ludhiana (Punjab). After that she completed her DNB (Pediatrics), then she did SIAMG fellowship in clinical genetics from Nizam's Institute of Medical Sciences (NIMS), Hyderabad, and PG certified course in medical and genetic counselling from Kamineni Hospital, Hyderabad.

Co Editor

Seema Kapoor MD (Pediatrics) is Director Professor, Division of Genetics, Department of Pediatrics, Maulana Azad Medical College, New Delhi. She has won many medals and awards during her graduation and postgraduation period. She has to her credit several articles published in journals of national and international fame and has contributed various chapters in different books. She is member of various ICMR research committees and committees related to health services for the community. She has played a significant role in getting achievements in the field of inborn errors of metabolism at the national level. She is also actively involved in teaching and academics and has been examiner for DM and DNB students in genetics.



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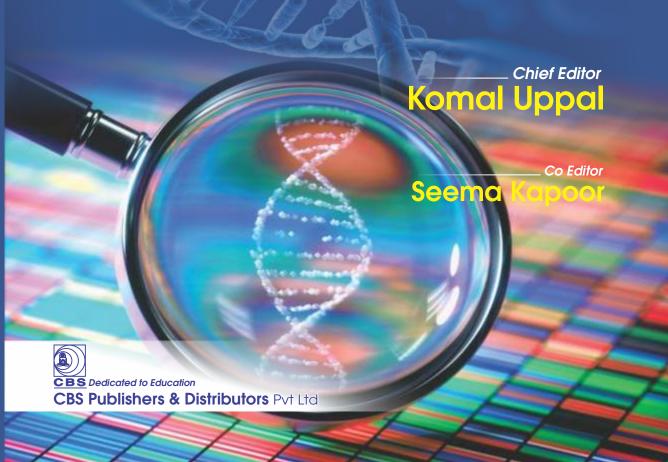


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for Students







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Chief Editor

Komal Uppal

DCH, DNB (Paediatrics), FSIAMG, PGCCMGC Medical Scientist Division of Genetics, AllMS, New Delhi

Co-Editor

Seema Kapoor

MD (Paediatrics)

Director Professor (Division of Genetics)

Department of Paediatrics

Maulana Azad Medical College, New Delhi

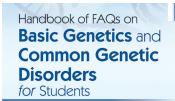


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4819/XI Prahlad Street, 24 Ansari Road, Daryaganj, New Delhi 110 002, India Ph: 011-23289259, 23266861, 23266867 Website: www.cbspd.com

Fax: 011-23243014 e-mail: delhi@cbspd.com; cbspubs@airtelmail.in

Corporate Office: 204 FIE, Industrial Area, Patparganj, Delhi 110 092, India

Ph: 011-4934 4934 Fax: 011-4934 4935 e-mail: publishing@cbspd.com; publicity@cbspd.com

Branches

 Bengaluru: Seema House 2975, 17th Cross, K.R. Road, Banasankari 2nd Stage, Bengaluru 560 070, Karnataka, India Ph: +91-80-26771678/79
 Fax: +91-80-26771680
 e-mail: bangalore@cbspd.com

• Chennai: 7, Subbaraya Street, Shenoy Nagar, Chennai 600 030, Tamil Nadu, India

Ph: +91-44-26680620, 26681266 Fax: +91-44-42032115 e-mail: chennai@cbspd.com

 Kochi: 42/1325, 1326, Power House Road, Opposite KSEB, Power House, Ernakulum-682018, Kochi, Kerala, India Ph: +91-484-4059061-67
 Fax: +91-484-4059065
 e-mail: kochi@cbspd.com

 Kolkata: 147, Hind Ceramics Compound, 1st Floor, Nilgunj Road, Belghoria, Kolkata 700056, West Bengal, India Ph: +91-9096713055/7798394118, 9836841399
 e-mail: kolkata@cbspd.com

Mumbai: PWD Shed, Gala No. 25/26, Ramchandra Bhatt Marg, Next to JJ Hospital Gate No. 2
 Opp. Uninon Bank of India, Noorbaug, Mumbai-400009, Maharashtra, India

Ph: +91-22-6661880/89 e-mail: mumbai@cbspd.com

Representatives

Hyderabad 0-9885175004
 Patna 0-9334159340
 Jharkhand 0-9811541605
 Nagpur 0-9421945513
 Pune 0-9623451994
 Uttarakhand 0-9716462459

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Contributors

Chief Editor

Komal Uppal

DCH, DNB (Paediatrics), FSIAMG, PGCCMGC Medical Scientist Division of Genetics, AIIMS, New Delhi email: uppalkomal3@gmail.com

Co-Editor

Seema Kapoor

MD (Paediatrics)

Director Professor (Division of Genetics)
Department of Paediatrics
Maulana Azad Medical College, New Delhi
email: drseemakapoor@gmail.com

Prajnya Ranganath

MD (Paediatrics), DM (Medical Genetics)
Associate Professor and Head
Department of Medical Genetics
Nizam's Institute of Medical Sciences
Hyderabad, Telangana
Adjunct Scientist
Diagnostics Division, Centre for DNA
Fingerprinting and Diagnostics, Hyderabad
email: prajnyaranganath@gmail.com

Inusha Panigrahi

MD (Paediatrics), Dip NB DM (Medical Genetics)
Professor
Genetic Metabolic Unit
Department of Paediatrics
APC, PGIMER, Chandigarh
email: panigrahi.inusha@pgimer.edu.in; inupan@yahoo.com

Neerja Gupta

MD, DM (Medical Genetics)
Associate Professor
Division of Genetics
Department of Paediatrics
AllMS, Ansari Nagar, New Delhi
email: neerja17@gmail.com; neerja17aiims@gmail.com

Kausik Mandal

MD (Paediatrics), DM (Medical Genetics)
Additional Professor
Medical Genetics
SGPGIMS, Lucknow
email: mandal.kausik@gmail.com

Deepti Saxena

MS (Obstetrics & Gynaecology), DM (Medical Genetics)
Assistant Professor
Department of Medical Genetics
Sanjay Gandhi Postgraduate
Institute of Medical Sciences
Raibareli Road, Lucknow
email: saxenadrdeepti@gmail.com

Chaitanya A Datar

MBBS, MD (Medical Genetics)
Clinical and Metabolic Geneticist
Bharati Hospital, Pune
and KEM Hospital, Pune
Surya Hospital and
Aditya Birla Memorial Hospital, Pune
email: dr.cdatar@gmail.com

Qurratulain Hasan

PhD, FNASc

Senior Consultant and Head Department of Genetics and Molecular Medicine Kamineni Hospitals, LB Nagar Hyderabad, Telangana email: qhasan 2013@gmail.com

Kuldeep Singh

MD (Paediatrics), DM (Medical Genetics)

Dean Academics

Head, School of Public Health and

Head, Department of Paediatrics

All India Institute of Medical Sciences, Jodhpur

email: kulpra@hotmail.com; kulpra@gmail.com

Usha R Dutta

PhD [Human Molecular Genetics], DAAD Fellow

Cytogeneticist

Diagnostics Division Centre for DNA

Fingerprinting and Diagnostics

(Ministry of Science & Technology, Gol)

Inner Ring Road, Uppal

Hyderabad, Telangana

email: usha@cdfd.org.inushadutta@hotmail.com

Seema Thakur

MS (Ob-Gyn), DM (Medical Genetics)

Senior Consultant

Genetic and Fetal Medicine

Fortis Hospital, Delhi-NCR

Rainbow Children Hospital, Delhi

email: Seematranjan@gmail.com

Meenakshi Lallar

MS (Ob-Gyn), DM (Medical Genetics)

Clinical Geneticist

Prime Imaging and Prenatal Diagnostics

Sector 24 D, Chandigarh

Consultant

Geneticist Medgenome, Bangalore

email: meenakshilallar@gmail.com

Divya Agarwal

MBBS, MD (Paediatrics), DM (Medical Genetics)

Consultant

Medical Genetics, Max Hospital

Gurgaon, Haryana

email: dr.divya2512@gmail.com

T Karthik Bharadwaj

MD (Paediatrics), DNB (Medical Genetics)

Principal

Clinical Geneticist

CSIR—Center for Cellular and Molecular Biology

Hyderabad, Telangana

email: karthikt@ccmb.res.in

Gayatri N

DGO, DNB (OBG), DNB (Medical Genetics)

Consultant

Medical Geneticist

Fernandez Hospital

Hyderabad, Telangana

email: drgayatri@gmail.com

Surya Balakrishnan

DNB (Internal Medicine), DNB (Medical Genetics)

Senior Clinical Geneticist

CCMB, Hyderabad

email: gierra@gmail.com

Mounika Endrakanti

MBBS, MD (Paediatrics), DM (Medical Genetics)

Senior Resident

All India Institute of Medical Sciences

New Delhi

email: mounika1404@gmail.com

Suresh Thomas MD (Paediatrics)

Professor and Head

Department of Paediatrics

Kamineni Institute of Medical Sciences

Narketpally, Nalgonda, Telangana

email: drsureshthomas@gmail.com

Shruti Bajaj MD (Paediatrics)

Fellowship Clinical Genetics (MUHS)

Consultant

Clinical Geneticist

Sir HN Reliance Foundation Hospital

Mumbai, Maharashtra

email: info@geneticsinindia.comdrshru.a@gmail.com

Sarah Bailur

MBBS, DNB (Paediatrics), FICG, FSIAMG

Paediatrician and Clinical Geneticist

Kamineni Hospitals, LB Nagar

Hyderabad, Telangana

email: medicosarah@gmail.com

Manisha Goyal

MBBS, DGO, Fellowship Medical Genetics and Fetal Medicine.

Postgraduate Diploma in Ultrasonography

Medical Geneticist

Centre of Rare Disease

JK Lone Hospital

SMS Medical College

Jaipur, Rajasthan

email: manidr2000@gmail.com

Snehal Patil MBBS, DCH

Fellowship Clinical Genetics (MUHS)

Senior Resident

SMBT Institute of Medical Sciences

and Research Center

Dhamangaon Igatpuri, Nasik, Maharashtra

email: sneh.doc@gmail.com

Ami Shah

DNB (Paediatrics), Fellowship Clinical Genetics (MUHS)

Consultant

Nanavati Super Specialty Hospital

Mumbai, Maharashtra

email: drami.rajesh.shah@gmail.com

Contributors

Suvarna Ghanashammagar

DNB (Paediatrics)

MGM Medical College

N-6, CIDCO, Aurangabad, Maharashtra email: drsuvarnamagar@gmail.com

orrain areavairrainagare gritaine orr

Risha Nahar Lulla

MSc (Med Gen), PhD (Gen Genomics)

Consultant, IBM Watson Health

Hyderabad, Telangana email: rishanahar@gmail.com

Syeda Zubeda

PhD (Genetics)

Scientific Officer

Department of Genetics

Kamineni Hospitals, LB Nagar

Hyderabad, Telangana

email: zubeda.syeda@yahoo.co.in

Priyanka Srivastava PhD

Assistant Professor

Genetic Metabolic Unit

Department of Paediatrics

Postgraduate Institute of Medical Education

and Research (PGIMER)

Sector-12, Chandigarh

email: srivastavapriy@gmail.com

Aruna Priya Kamireddy

MSc, Human Genetics [PhD]

Research Scholar

Kamineni Hospitals

LB Nagar, Hyderabad

email: aru.priya10@gmail.com

Gayatri R Iyer

PhD

Human Genetics and Senior Research Fellow

DST INSPIRE Fellow

Kamineni Hospitals

Hyderabad, Telangana

email: gaya3riyer@gmail.com

Varuna Vyas

MBBS, MD (Paediatric Endocrinology)

Assistant Professor

AllMS, Jodhour, Raiasthan

email: drvaruna.vyas@gmail.com

Soma Santosh Kumar DCH, DNB (Paediatrics)

Assistant Professor

Department of Paediatrics

Kamineni Institute of Medical Sciences

Narketpally, Nalgonda, Telangana

email: santoshsoma880@gmail.com

Meenakshi Bothra Gupta

MD (Paediatrics)

Assistant Professor

Department of Paediatrics

Maulana Azad Medical College

email: meenakshibothra@gmail.com

Siyaram Didel MD (Paediatrics)

Assistant Professor

AIIMS, Jodhpur, Raiasthan

email: drdsram2001@gmail.com, didelsr@aiimsjodhpur.edu.in

Shreya Tanneru

MBBS, MD (Paediatrics)

Assistant Professor

Department of Paediatrics

Kamineni Institute of Medical Sciences (KIMS)

Nalgonda, Telangana

email: drshreyatanneru@gmail.com

Irene Mathews

MD (Dermatology, Venereology and Leprology), DNB, SCE

Post-Doctoral Fellowship in Paediatric Dermatology

JIPMER, Puducherry

email: irenemathews7@gmail.com

Jerene Mathews

MD, Fellowship (Paediatric Dermatology)

Assistant Professor

Department of Dermatology

Believers Church Medical College

Thiruvalla, Kerala

email: jerenemathews7@gmail.com

Yashodhara Bhattacharya MSc (Biomedical Genetics),

Post Graduate Diploma in Medical and Genetic Counseling

Genetic Counselor

Redcliffe Lifesciences

Kolkata, West Bengal

email: y.bhattacharya12@gmail.com

Divya Kumari

Senior Research Fellow (SRF)

Genetic Metabolic Unit

Department of Paediatrics

APC, PGIMER, Chandigarh

email: kumari.divya093@gmail.com

Foreword

t gives me great pleasure to provide this introduction to the first editon of the *Handbook of FAQs on Basic Genetics and Common Genetic Disorders for Students*. This book is a unique compilation of important concepts related to medical genetics in a question and answer format. Though the primary intent behind the effort is to provide a ready reference to undergraduate and postgraduate medical students to help them attempt questions related to medical genetics in examinations, it also serves the much bigger purpose of elucidating the principles and practice of medical genetics to medical students at an early stage in their career.

Introducing the concepts of medical genetics and highlighting the relevance of this subject to medical practice to students, right from their undergraduate training period is imperative in this field of genetic diagnostics and molecular therapy in recent years. Apart from the so-called 'pure genetic disorders' which are known to be caused by chromosomal abnormalities and genetic mutations, a growing number of multifactorial disorders including diabetes, hypertension, malignancies, autoimmune diseases, psychiatric disorders and many others, are being identified to have genetic

contributory factors in their causation. These chromosomal disorders and monogenic diseases, as well as multifactorial disorders can affect all body organs and organ systems. Therefore, all medical practitioners, irrespective of their field of specialization, need to understand the basic conceps of genetics as well as be aware about these recent advances.

Unfortunately, at present, very little emphasis being given to genetics in undergraduate medical teaching and postgraduate medical training. Though genetics is a part of the medical curriculum, it is often mistakenly considered to be a non-clinical research area, not relevant to clinical practice. Students also get put-off by the so-called 'complex terminology' and 'hard-to-grasp' concepts of medical genetics.

This book is an attempt to simplify the concepts of genetics, highlight the importance and relevance of the subject in medical practice, and inculcate an interest in the subject in medicos early in their career. I congratulate the editorial team for this wonderful and timely effort. I am sure this book would prove to be of great help to medical students not just in their estimations but also in their practice.

Dr Prajnya Ranganath

MD (Paediatrics), DM (Medical Genetics)
Associate Professor and Head
Department of Medical Genetics
Nizam's Institute of Medical Sciences
Hyderabad

Preface

enetics is an essential subject in the field of human science. In the last two decades due to various advancements in diagnostics and therapeutic genomic techniques, the whole world has got transformed. But the medical students are still lagging behind in understanding the fundamental concepts of genetics and common genetic disorders. Even though the chapters on the basic information related to genetics are introduced at the middle school level but still the foundational knowledge of the medical students about genetics is not satisfactory.

Because of this reason, students are not very comfortable in writing the answers related to genetics in the examinations.

In view of the above, via this book, an attempt is made to bridge the gap between the low level and the expected level of knowledge of students about the subject and to facilitate them to develop the required knowledge base, and skills to write correct answers in the examination. This book will be having a

wide variety of questions and their answers based on basic cellular and molecular genetics, common genetic disorders, conventional and advanced diagnostic techniques, treatment and prevention of genetic disorders and recent advances in the genetics. To prepare these questions and answers various expert contributors, some of whom being eminent teaching faculty in medical colleges, with their vast knowledge about the subject and understanding the needs of the students, have spent a lot of time and energy.

With the objective of providing comprehensive and basic information on genetics—almost all the FAQs related to genetics which are frequently asked in the examinations have been covered in a simple and easy language in this book. Hopefully, the book will be friendly and a single window solution for students to revise the subject before the examinations.

Any suggestion to improve the book will be greatly acknowledged.

Komal Uppal

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- Co-editor, Dr Seema Kapoor, without her contributions this book would not have come to being.
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I have made all the attempts to acknowledge all the sources of information and relevant illustrations. If something has left out unintentionally, I apologize for that.

Komal Uppal

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