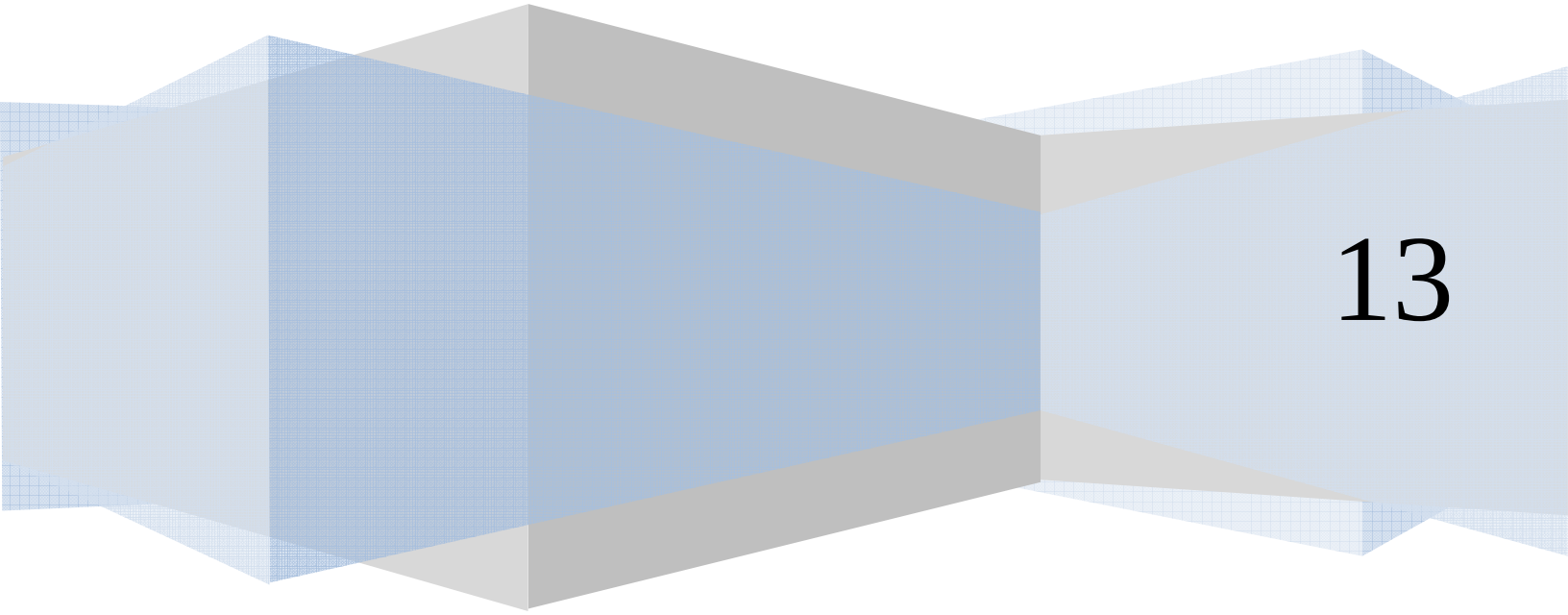
Genetics Division
Department of Pediatrics
KAMC-RD, NGHA

# ORIENTATION BOOKLET FOR RESIDENTS AND FELLOWS

## ROTATING IN GENETICS SERVICE AT KING ABDULAZIZ MEDICAL CITY



13

# ORIENTATION BOOKLET FOR RESIDENTS AND FELLOWS DURING THEIR ROTATION IN GENETICS SERVICE AT KING ABDULAZIZ MEDICAL CITY

## Table of Contents

## I. INTRODUCTION

Medical Genetics is a branch of medicine concerning the effects of genetic variations on human development and health. It is also the study, diagnosis, management, and prevention of genetic and related disorders in individuals, families and communities.

The Genetics division in King Abdulaziz Medical City is currently subdivided into two sections:

- **Clinical Genetics:** caring mainly about patients with dysmorphism, clinical syndromes, cancer genetics and chromosomal abnormalities.
- **Biochemical Genetics (Metabolic):** caring mainly about patients with inborn errors of metabolism.

Your rotation will be divided into two parts, (1st half with clinical genetics, and 2nd half with biochemical genetics) or vice versa. The genetics secretary will give you orientation booklet, surveys to be filled up and your schedule prior to the start of your rotation.

## II. FACULTY:

**Consultants:**

- Dr. Majid Alfadhel
- Dr. Abdulrahman Al Swaid
- Dr. Wafaa Eyaid

**Associate Consultant:**

- Dr. Faroug Ababneh

**Assistant Consultant:**

- Dr. Abdulrahman Obaid

**Coordinators:**

- Rasha AlKendi
- Hanan Alfifi
- Omniah Abdulrehim

**Secretary:**

- Daisy Ancheta

## III. OBJECTIVES:

As basic genetic knowledge continues to be integrated into medical practice, it is important to train all clinicians in some aspects of medical genetics.

In this rotation, we will offer you some training in patient examination, diagnosis, decision making, therapeutic and counseling required to provide care for patient with genetic or possible genetic disease.

Additionally, you will learn about the usefulness and limitation of lab tests used in the diagnosis and genetic disorders.

As a Resident/Fellow rotating in Medical Genetics, you will learn the approach of management of patient with:

- Congenital Anomalies and Dysmorphic Features
- Genetic Counseling
- Basic Genetic Terminologies
- Family pedigree and mode of inheritance
- Management of Inborn Errors of Metabolism Crises and long term follow up of common metabolic disorders
- Clinical genetics approach to various genetics clinical presentations

## IV. CURRICULUM:

| DAY | 8:00-8:30 | 8:30-12:00 | 12:30-13:00 | 13:00-17:00 |
|---|---|---|---|---|
| **Thursday** | Morning report | In patient /Consultation | | Clinical Genetics Clinic<br>Dr.Swaid |
| **Sunday** | Morning report | Clinical Genetics Clinic<br>Dr.Swaid<br>Biochemical Genetics Clinic<br>Dr. Alfadhel | | Biochemical Genetics Clinic<br>Dr.Eyaid<br>Dr. Alfadhel |
| **Monday** | Morning report | Monitoring dietitians Round (9-10)<br>Inpatient /Consultation | | Inpatient /Consultation |

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|---|---|---|---|---|
| **Tuesday** | Morning report | Biochemical Genetics Clinic<br>Dr.Eyaid | Journal Club | Inpatient /Consultation |
| **Wednesday** | Morning report | Inpatient /Consultation | | Grand Round |

## V. TOPICS TO BE DISCUSSED DURING ROTATION:

1. Approach of dysmorphic child and common dysmorphic terminology.
2. Approach of newborn/infant with metabolic crises.
3. How to write and read Family Pedigree and analyze the mode of inheritance.
4. The basic genetic terminology.
5. Classification of metabolic disorders and common presentation
6. How to search the Oxford Medical database and Possum Medical database.
7. Medical literature search skills.
8. Management of acute metabolic crises
9. Clinical approach to various genetics presentations
10. Case scenarios of different Inborn Errors of Metabolism

The discussion will be mainly depending on your preparation and degree of seeking the information. The consultants will focus mainly on the patient based approach, however, if you wish to discuss additional topics, then, you should ask this directly from the appropriate consultant and will save time for you to discuss these topics.

## VI. EXPECTATIONS FROM YOU DURING ROTATION:

1. Punctuality in attending the morning report, the clinic and grand round.
2. Punctuality in doing consultation.
3. Taking care of inpatients daily.
4. Comprehensive brief daily notes for your patient.
5. Clark the consultation form and search about the possible diagnosis and presenting it to your staff in a comprehensive way.
6. Present a case you see during your rotation and review literature about this case.
7. **Consultation Service:**
    - Routine consultation will be done on a “first come, first served” basis. However, the priority is for more urgent cases (i.e. NICU/PICU).
    - **Pedigree form should be filled for every consultation unless done before and needs only update.**
    - Follow up on acute consultations. Plan for follow up prior to discharge.
    - Follow up the laboratory investigations results and if it was done or not
    - Each consultation should be copied and handed to secretary/coordinator to be uploaded in genetics shared folder data base.
    - There are genetics folder available at all clinical wards and outpatient contains all requisitions forms specific for genetics service and also family pedigree form.

**You are expected to write a detailed note during the first encounter with the patient. The notes should include, but not be restricted to the following:**

- Reason for consultation
- Pregnancy history (fetal movement, U/S reports, history of poly- or oligohydramnios, history of gestational diabetes or pre-eclampsia, history of infections, premature rupture of membrane or maternal fever).
- Delivery history (birth weight, length, head circumference, apgar scores, place of delivery).
- Neonatal history (numbers of NICU admission, neonatal jaundice, any problem during neonatal period).
- Nutritional history.
- Detailed history about current problems and when the first concern was noted.
- Developmental history.
- 3-Generation family history and history of consanguinity and ethnicity of the parents.
- If relevant, social history and vaccinations history.
- Examination: should include: growth parameters (height, weight, head circumference), description of dysmorphic features (if present), detailed examination of the most affected system, brief examination of other system
- Investigations done for the patients.

## VII. INPATIENT (BIOCHEMICAL GENETICS) SERVICE:

A. Daily note about admitted patient, the note should be in the form of SOAP (S: subjective, O: objective, A: assessment, P: plan). The note should contain but not restricted to the following:
    - Detailed nutritional history, total caloric intake, total protein intake (natural protein eg: similac formula and synthetic protein eg: propiomex formula)
    - Medications and dosages/kg/day
    - Input-output and fluid balance

B. Every Tuesday: there are metabolic patients coming to Day Care Unit for Enzyme Replacement Therapy. You are expected to see those patients, write a brief note and help the day care staff for any issue raised during their stay.

C. Several biochemical genetics patients need skin biopsy for C/S fibroblasts in order to do various genetics testing or sometime lumbar Puncture for CSF neurotransmitters. It would be preferred if you could learn such procedures and train yourself on it during your rotation

D. The protocols for management of common Inborn Errors of Metabolism are available at all clinical wards, ER, PICU and NICU.

E. Some biochemical genetics patients came to outpatient clinic for heparin flush of their central line. You are expected to manage any issue raised during their stay like ordering heparin flush for example.

## VIII. LEAVE:

In general, because your rotation in genetics service is too short, the leaves are not allowed.

## IX. PERMISSION TO BE ABSENT FOR ONE OR TWO DAYS DURING YOUR ROTATION DUE TO SOCIAL REASONS:

In general, we do not encourage you to ask such requests however, this should be discussed with the consultant who is on clinical duty during your proposed leave.

## X. CONTACT NUMBERS:

| | PHONE | PAGER |
|---|---|---|
| Dr. Majid Alfadhel | 12596 | 3069 |
| Dr. Abdulrahman Al Swaid | | 5625 |
| Dr. Wafaa Eyaid | 12282 | 8171 |
| Dr. Faroug Ababneh | | 1386 |
| Rasha Alkendi | 12513 | 1137 |
| Hanan Alfifi | 11426 | 2306 |
| Omniah Abdulraheem | 11707 | 1989 |
| | | |
| | | |
| Cytogenetic Lab<br>Dr. Esam Amir | 11360 | 8211 |
| Research Lab<br>Dr. Abdulkarim | 16665/16663 | 6664 |
| Molecular Genetics lab<br>Dr. Mohammed Albalwi | 11680/11287 | 1287 |
| Biochemical metabolic lab<br>Dr. Ali Alothaim | 40178/40196/40195 | |
| | | |
| King Faisal Specialist Hospital | 0114647272 | |
| Metabolic lab in KFSH | 32695/32635 | |
| Receiving/send out Lab | 11174/11176 | |
| Sent out lab (Shirley) | 13261 | |
| **Biochemistry supervisor**<br>Dr. Waleed Tamimi | 11899<br>8212 | |
| X-ray film | 11317/11413 | |

| | | |
|---|---|---|
| Medical Record (chart review) | 18759<br>7687 | |
| **Muscle Pathologist**<br>Dr. Hider | 12145<br>1236 | |
| **Pediatric ophthalmologist** | | |
| Dr Saadon | 8886 | |

## XI. Suggested Readings

- **Resident suggested Books:**

1) Nelson Textbook of pediatrics
2) Genetics in Medicine, Thomson &Thomson , M.Innes
3) A Clinical Guide to Inherited Metabolic Disease, J.T.R Clarke
4) Atlas of Common Lysosomal and Peroxisomal Disorders, Pinar T. Ozand and Mohamed Al Essa
5) Recognizable Patterns of Human Malformation, K. Lyon ones 1988 (Revised edition of "Smith"). A good book but not the only book for dysmorphology.
6) Handbook of Normal Physical Measurements, Hall, J.G., Froster-Iskenius, U.G., and Allanson, J.E. 1989.
7) Vademecum Metabolicum, Zschocke/Hoffmann
8) Medical Genetics, I an D. Young
9) Inborn Metabolic disease, Diagnosis and Treatment. Saudubray, Van den Berghe, Walter

- **Fellow's suggested Books**

10) **Recognizable Patterns of Human Deformations**, J.M. Graham 1988.

11) **Syndromes of the Head and Neck**, Gorlin R.J., Cohen M.M. Jr, Levin L.S. 3rd edition 1990. Oxford University Press. A very useful book for all patients.

12) **Bone Dysplasias, An Atlas of Constitutional Disorders of Skeletal Development**, Sprang, Langer and Widemann, 1974. The ultimate source for x-ray findings. Out of print.

13) **Mendelian Inheritance in Man**, V.A. McKusick, 1994. Capsule summaries with good references. Available on-line in Blalock 10 and in Welch as OMIM.

14) **The Malformed Infant and Child**, Goodman and Golin, 1983. Slightly different point of view than #3. Easier to carry.

15) **Birth Defects Encyclopedia**, Boyese, Mary Louise editor-in-chief 1990. Good, recent, quick reference.

16) **Human Malformations and Related Anomalies**, Stevenson R.E., Hall J.G., and Goodman R.M. 1993. Oxford University Press. A useful guide in terms of differential diagnosis.

17) **Heritable Disorders of Connective Tissue**, V.A. McKusick, 1994. For all connective tissue disorder.

18) **Genetics and Hand Malformations**, Temtamy and McKusick. Expensive but good for these problems.

19) **Atlas of Mental Retardation**, L. Holms. Expensive but good pictures and text.

20) **Human Cytogenetics**, Vol. 1&2, J. Hamerton, 1971. Good basic cytogenetic text.

21) **Clinical Atlas of Human Chromosomal Disorders**, de Grouchy and Turleau, 1984. Good clinic-karyotype correlation.

22) **New Chromosomal Syndromes**, J. Yunis, 1977. New techniques and syndromes.

**23) Genetic counseling aids, green wood genetic centre**

**24) Inherited metabolic diseases, George F. Hoffmann, Johannes Zschocke, William Nyhan**

## XII. Web site to visit during the rotation:

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| http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM | ***OMIM - Online Mendelian Inheritance in Man*** |
| http://learn.genetics.utah.edu/ | ***Learn. genetics*** |
| http://www.ncbi.nlm.nih.gov/gtr/ | ***Gene tests*** |
| http://www.genome.gov | ***National Human Genome Research Institute*** |
| http://www.ac-knowledge.net/ngha/ | ***NGHA library*** |
| www.pubmed.com | ***Pubmed*** |
| http://www.google.com.sa/ | **Google** |
| http://www.genetests.org/ | ***Genereview*** |

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|---|---|
| http://www.kumc.edu/gec/geneinfo.html | ***Information for genetic professionals, University of Kansas Medical Center*** |
| http://www.faseb.org/genetics/acmg | ***American College of Medical Genetics*** |
| http://www.abmg.org/ | ***American Board of Medical Genetics*** |
| http://www.ccmg-ccgm.org/ | ***Canadian College of Medical Geneticists*** |
| http://www.ashg.org/ | ***American Society of Human Genetics*** |
| http://www.ashg.org/genetics/ | ***A World of Genetics Societies*** |
| http://www.eshg.org/ | ***European Society of Human Genetics*** |
| http://www.geneticalliance.org/ | ***Genetic Alliance*** |
| http://www.ensembl.org./ | ***Information on Genomics*** |
| http://www.reprotox.org/ | ***Information on Teratology*** |
| http://genetics.ich.ucl.ac.uk | ***London Dysmorphology Discussion Board*** |
| http://rarediseases.info.nih.gov | ***Office of Rare Diseases*** |
| http://www.ssiem.org/ | ***Society for the Study of Inborn Errors of Metabolism*** |
| http://www.nchpeg.org/ | ***The National Coalition for Health Professional Education in Genetics*** |

**Best wishes for you during your rotation with us**