Module 3, BSc in Haematology 2012-2013

Inherited and acquired red cell disorders

**Learning objectives and timetable**

Module convenors:

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At the end of this module the student should be able to

* Describe normal and abnormal haemopoiesis
* Describe the structure and function of haemoglobin A and explain how haemoglobins A2 and F differ from haemoglobin A
* Describe the alpha and beta globin gene clusters and explain the control of globin chain synthesis
* Explain what is meant by the terms: beta thalassaemia, beta0 thalassaemia, beta+ thalassaemia
* Describe the clinicopathological features of beta thalassaemia minor, beta thalassaemia intermedia and beta thalassaemia major and relate these terms to the possible underlying genetic defects
* Describe the significance of beta thalassaemia to individual patients and their families
* Diagnose beta thalassaemia trait and explain how other thalassaemia syndromes are diagnosed (including explaining the principles of molecular diagnostic techniques)
* Explain the terms: alpha+ thalassaemia and alpha0 thalassaemia and describe how they interact to cause haemoglobin H disease and haemoglobin Bart’s hydrops fetalis; describe the clinicopathological features of these alpha thalassaemia syndromes
* Describe the clinicopathological features of common haemoglobinopathies including haemoglobin S (covered in introduction to BSc course), haemoglobin C and haemoglobin E
* Explain how mutations in globin genes can cause functional abnormalities in haemoglobin (high or low affinity haemoglobins, methaemoglobinaemia, unstable haemoglobins)
* Explain how thalassaemias interact with haemoglobinopathies
* Discuss how antenatal and neonatal screening for haemoglobinopathies are carried out
* Describe inherited and acquired bone marrow failure syndromes and discuss their diagnosis
* Describe the structure of the red cell membrane and explain how inherited defects of the red cell membrane lead to haemolytic anaemia
* Describe the enzyme pathways that are important in red cells
* Explain why the pentose shunt is important and explain the causes and effects of deficiency of glucose-6-phosphate dehydrogenase (G6PD)
* Summarise the inherited causes of haemolytic anaemias according to pathogenesis
* Discuss the effects of malaria on the red cell genome
* Discuss the prospects for gene therapy for inherited red cell disorders

**Supplementary Reading (available in library).**

***An up-to-date basic book:***

Bain BJ. Haematology: a Core Curriculum. Imperial College Press, London, 2010.

***A useful reference book:***

Hoffbrand AV, Catovsky D, Tuddenham EGD and Green AR (Eds) Postgraduate Haematology, 6th Edition, Wiley–Blackwell, Oxford, 2010.

***Useful for this module***:

Abdalla SA and Pavsol G (Eds) Malaria: a Hematological Perspective. Imperial College Press, London, 2004.

Bain BJ, Haemoglobinopathy Diagnosis, Blackwell Publishing, 2nd Edn, Oxford, 2006. Also available electronically at: <http://www3.interscience.wiley.com/cgi-bin/bookhome/116839557>

Weatherall D and Clegg JB. *The Thalassaemia Syndromes*. 4th Edn., Blackwell Science, Oxford, 2001.

Steinberg MH, Forget BG, Higgs DR and Weatherall DJ (eds) *Disorders of Hemoglobin: Genetics, Pathophysiology and Clinical Management*, 2nd Edn, Cambridge University Press, Cambridge, 2009.

Knight JC. Human Genetic Diversity. Oxford University Press, Oxford, 2009.

**Note to teachers (except external guest lecturers)**

For each lecture, the students should either receive a handout (1-2 pages of A4) or have access to a Power Point Presentation (PPP) (or both). Either a handout or a PPP should be sent to webmaster.feo@imperial.ac.uk so that it can be placed on the intranet some days in advance of your lecture. Some students like to read the PPP in advance so that they can then understand the lecture better and can print it in advance and use the printout to take notes. If your PPP is too large a file to email it should be sent to the webmaster through <https://fileexchange.imperial.ac.uk> and the webmaster should be notified by email of its location.

If external guest lecturers are willing to leave a copy of a PPP for the benefit of the students this would be greatly appreciated as it saves the students having to take detailed notes.

**Week 1 - St Mary’s Campus**

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| **Monday 31st****December** | **Tuesday 1stJanuary 2013** | **Wednesday 2nd January** | **Thursday 3rd January** | **Friday 4th January** |
|  | PUBLIC HOLIDAY  | 09.00-10.00**Dr A Porter**Gene structure and function, Globin genes (revision)**Cockburn Lecture Theatre**  |  | 09.00-10.00**Dr S Abdalla**Introduction to module &epidemiology of inherited red cell disorders**Anthony De Rothschild Lecture Theatre** |
| 10.00-11.00**Professor B Bain**Sickle cell disease (revision)**Cockburn Lecture Theatre**  | 10.00-11.00**Professor B Bain**β thalassaemia (lecture + video) **Peart Room-QEQM** | 10.00-11.00**Professor B Bain**α thalassaemia **Anthony De Rothschild Lecture Theatre** |
| 11.00-12.00**Professor B Bain** Haemoglobin Structure and Function (Revision) **Cockburn Lecture Theatre** | 11.00 – 12.00**Professor B Bain**The interaction of β thalassaemia with HbS and HbE **Peart Room-QEQM** | 11.00 – 12.00**Professor B Bain** Living with thalassaemia major DVD **Anthony De Rothschild Lecture Theatre** |
| 12.00-13.00**Mr D Roper**Variant haemoglobins **Cockburn Lecture Theatre** | 12.00-13.00**Professor B Bain**CAL 1st session (thalassaemias)**Hynds Computer Lab**  | 12.45-13.45**STAFF ROUND****Cockburn Lecture Theatre** |
| 13.00-14.00 **Professor B Bain**CAL 2nd session for those not attending first session**Hynds Computer Lab** |
| SPORTS AFTERNOON | 14.00-14.30**Ms L Phelan and Professor B Bain**First half of group: Visit Diagnostic Lab, First Floor, old building) half group **Cockburn Lecture Theatre** | 14.00-15.00**Dr S Abdalla** Malaria: clinical aspects and pathophysiology**Peart Room-QEQM** |
| 14.30 -15.00**Ms L Phelan and Professor B Bain**Second half of group: Visit Diagnostic Lab, First Floor, old building) **Cockburn Lecture Theatre** |  |

**Week 2 – Monday Hammersmith Campus; Tuesday – Friday St Mary’s Campus**

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| **Monday 7th January Hammersmith**  | **Tuesday 8th January** | **Wednesday 9th January** | **Thursday 10th January** | **Friday 11th January** |
| 09.30-10.30**Professor I Roberts**Normal Haemopoiesis**Wolfson Education Centre Seminar Room 2** | 09.15-11.00 Possibility of attending clinic (no more than 2 students with Dr Abdalla and his SpR and another 2 with Dr Rowley and her SpR)  | OPD Clinic (no more than 2 students with Dr Abdalla and his SpR and another 2 with Dr Shlebak and his SpR)**Clinic E, OPD** | 10.00 to 11.00**Dr J de la Fuente**The role of stem cell transplantation in the haemoglobindisorders **Peart Room-QEQM** | OPD Clinic (no more than 2 students with Dr Marks and her SpR and another 2 with Dr Layton and his SpR)**Clinic D, OPD** |
| 10:30-11:30**Professor I Roberts**Haemoglobinopathies in neonates**Wolfson Education Centre Seminar Room 2** | 11.00 – 12.00**Professor B Bain**Scientific Writing and how to give a power point presentation **Peart Room-QEQM** |
| 11.00-12.00**Dr Helen New**Overview of red cell enzymes**Clinical Lecture Theatre** |
| 11.30-12.30 **Prof Letizia Foroni** Revision Seminar: molecular techniques in red cell disorders**Wolfson Education Centre Seminar Room 2** | 12.30-13.30 **Guest lecturer****Dr Stephen Field**Indications and contraindications for transfusion in inherited red cell disorders**Peart Room-QEQM** |  | 12.30-13.30**Guest lecturer** **Dr Noemi Roy** The alpha thalassaemias**Cockburn Lecture Theatre** | 12.45-13.45**STAFF ROUND****Cockburn Lecture Theatre** |
| 13.00-15.00**Dr Carolyn Millar and Dr Andi Roy**Microscopy (thalassaemias and variant haemoglobins) **Southside Lab 7S2**Level 7 Commonwealth Building | 14.00-15.00**Guest lecturer****Dr Stephen Field**Transfusion in the Developing World**Peart Room-QEQM** | SPORTS AFTERNOON | 14.00-15.00Developing an antenatal diagnostic service**Ms Lori Phelan****Professor B Bain** **Cockburn Lecture Theatre**  | PRIVATE STUDY (preparation of power point presentation) |
| 15.00-15.30**Mr David Roper** Practical demonstration **Southside Lab 7S2**Level Commonwealth Building | 15.00-16.00 **Guest lecturer****Dr Stephen Field** AIHA and CHAD **Cockburn Lecture Theatre** | 15.00 – 17.00**Professor B Bain** HaemoglobinopathyPractical (sheets) **Cockburn Lecture Theatre** |
| 15.45-17.00**Dr D Marin**Workshop: Interpreting survival curves**This is the re-scheduled session from 16/11/12** CWB Library Training Room Commonwealth Building | 16.00-17.00**Dr Saad Abdalla**The Spleen**Cockburn Lecture Theatre** |

**Week 3 – Monday Hammersmith Campus; Tuesday – Friday St Mary’s Campus**

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| **Monday****14th January****Hammersmith** | **Tuesday 15th January** | **Wednesday 16th January** | **Thursday 17th January** | **Friday 18th January** |
| 10.00-11.00 **Professor B Bain**CAL Haemolytic anaemias**Wolfson Education Centre Lecture Theatre 1** | Possibility of attending clinic (Dr Abdalla and Dr Rowley)  | OPD Clinic (no more than 2 students with Dr Abdalla and his SpR and another 2 with Dr Shlebak and his SpR)**Clinic E, OPD** | Possibility of attending lymphoma clinic (Dr Marks) | OPD Clinic (no more than 2 students with each consultant)**Clinic D, OPD** |
| 11.00 -12.00  **Dr Carolyn Millar**  **and Dr Andi Roy** Haemolytic anaemias (microscopy) **Wolfson Education Centre Seminar Room 3** |
| 13:00-14:00**Dr K Anie** Psychosocial aspects of SCD**Wolfson Education Centre Seminar Room 4** | 11.30-12.30**Guest Lecturer****Dr Anne Yardumian**Beta thalassaemia intermedia **Cockburn Lecture Theatre** |  |   | 12.45-13.45**STAFF ROUND****Cockburn Lecture Theatre** |
|  |  | SPORTS AFTERNOON |  |  |
| 14.00-15.00**Prof T Karadimitris**Paroxysmal nocturnal haemo-globinuria**Wolfson Education Centre Seminar Room 4** | 14.00-15.00 **Dr S Chakravorty**Red cell membrane and its defects**Cockburn Lecture Theatre** | PRIVATE STUDY(preparation of power point presentation) | PRIVATE STUDY (preparation of power point presentation) |
| 15.00 – 16.00**Prof T Karadimitris**Haemophagocytic lymphohistiocytosis**Wolfson Education Centre Seminar Room 4** | 15.00 to 16.00**Dr S Abdalla**Infectious causes of bone marrow dysfunction**Cockburn Lecture Theatre** |
| 16.00-17.00**Prof Letizia Foroni**Revision: clarifying difficult concepts**Wolfson Education Centre Seminar Room 4** |  |

**Week 4 - Monday Hammersmith Campus; Tuesday – Friday St Mary’s Campus**

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| **Monday 21st January****Hammersmith** | **Tuesday 22nd January** | **Wednesday 23rd January** | **Thursday 24th January** | **Friday 25th January** |
|  | 09.15 – 10.00**Dr S Chakravorty** Discussion of sickle cell disease with a patient**Cockburn Lecture Theatre** | 09.15-12.00OPD Clinic (no more than 2 students with Dr Abdalla and his SpR and another 2 with Dr Shlebak and his SpR)Clinic E, OPD | 09.00-10.00 **Dr J de la Fuente**Inherited and acquired red cell aplasia **Anthony De Rothschild Lecture Theatre** | 09.00-12.30**Dr Saad Abdalla and Dr Nichola Cooper****IN COURSE ASSESSMENT****POWER POINT PRESENTATIONS****Antony De Rothschild Lecture Theatre** |
| 10.00-11.00**Professor Marina Botto**Complement **Wolfson Education Centre Seminar Room 2** | 10.00-11.00**Dr Helen New**G6PD deficiency**Cockburn Lecture Theatre**  | 10.00-11.00 **Dr J de la Fuente**Congenital bone marrow failure syndromes**Anthony De Rothschild Lecture Theatre** |
| 11.00-12.00**Dr A Porter**Prospects of gene therapy in haematological disorders**Wolfson Education Centre Seminar Room 2** | 11.00-12.00**Dr Saad Abdalla**Hyperreactive Malarial Splenomegaly**Cockburn Lecture Theatre** | 11.00-12.00 **Dr Mark Layton** Diagnostic problem solving in thalassaemia**Anthony De Rothschild Lecture Theatre** |
|  |  | **SPORTS AFTERNOON** | 12.15-13.45**Dr Mark Layton** Historical account of the discovery of red cell disorders**Anthony De Rothschild Lecture Theatre** | 12.45-13.45**STAFF ROUND****Cockburn Lecture Theatre**  |
| 14.00-15.00**Dr Mark Layton**Globin chain disorders and their treatment**Wolfson Education Centre Lecture Theatre 3**  |  |  | 14.00-17.00**Dr Saad Abdalla and Dr Nichola Cooper****IN COURSE ASSESSMENT****POWER POINT PRESENTATIONS****Anthony De Rothschild Lecture Theatre**  |
| 15.00-16.00 **Dr Mark Layton**Inherited haemolytic anaemias**Wolfson Education Centre Lecture Theatre 3** |

**Week 5 - St Mary’s Campus**

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| **Monday 28th January** | **Tuesday 29th January** | **Wednesday 30th January** | **Thursday 31st January** | **Friday 1st February** |
| PRIVATE STUDY (essay writing) | **ESSAYS TO BE SUBMITTED ELECTRONICALLY VIA BLACKBOARD BY 22.00 HRS** 10.00-12.00 Possibility of attending clinic (Dr Abdalla and Dr Rowley) | OPD Clinic (no more than 2 students with Dr Abdalla and his SpR and another 2 with Dr Shlebak and his SpR)Clinic E, OPD | 10.00 -11.00 **Prof Letizia Foroni**Revision tutorial**Cockburn Lecture Theatre**  | OPD Clinic (no more than two students with each consultant)**Clinic D, OPD** |
| 11.00-12.00**Dr Stephen Marks** Revision cases**Roger Bannister Lecture Theatre** |
| PRIVATE STUDY (essay writing) | 12.30-13.30**Guest Lecturer:** **Dr David Rees**Hb E **Anthony De Rothschild Lecture Theatre** | **SPORTS AFTERNOON** |  | 12.45-13.45STAFF ROUND **12.30-14.00****Cockburn Lecture Theatre** |
| PRIVATE STUDY (essay writing) | 14.00-15.00**Dr S Abdalla**Revision tutorial and feedback on course including Power Point presentations and Essays**Clinical Lecture Theatre** |  |  |

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