PLENARIES

PLENARY 1 (PL1): Global Burden of Haemoglobin Disorders

Professor David C Rees

David Rees is Professor of Paediatric Sickle Cell Disease at King's College, London. He studied medicine at Cambridge University and St Thomas' Hospital London. He started haematology training at University College Hospital, London where he developed an interest in sickle cell disease and thalassaemia. In Oxford, he was involved in research into causes of phenotypic variability in patients with HbE/b thalassaemia under the supervision of Professor Sir David Weatherall and Professor John Clegg.

At King's College Hospital, he has developed an active clinical and laboratory research programme in children with sickle cell disease, which won the BMJ Secondary Care Team of the Year award in 2011. He has also established a centre for acute porphyria at King's College Hospital, one of only two in the UK. With his network of centres for children with inherited anaemia across south London and southern England, he is involved in the care of up to 1000 children with these conditions. He is a medical adviser to the Sickle Cell Society, a member of Sickle Cell and Thalassaemia Screening Committee and a medical adviser to the Sickle Cell and Thalassaemia Screening Programme for England.

SUMMARY

Objectives:

1. To describe the world distribution of haemoglobin disorders
2. To describe the health burden of haemoglobin disorders

Haemoglobin disorders are the commonest, severe inherited disorders in the world, particularly prevalent in low- and middle-income countries. The two main conditions caused by mutations in globin genes are sickle cell disease (SCD), and thalassaemias.

Approximately 300 000 children are born each year with sickle cell disease, with estimates suggesting this will reach 400 000 per year by 2050. The largest numbers of SCD births are estimated to occur in Nigeria (90 000 births/year), Democratic Republic of Congo (39 700 births/year) and India (44 000 births/year). Worldwide there are approximately 30 000 births/year of babies with thalassaemia major, and 20 000 with HbE/□ thalassaemia, with the most of these occurring in Asia and the Middle East.

The relative importance of haemoglobin disorders is increasing in many countries as deaths from infectious diseases decrease, and both thalassaemia and SCD are recognised as global health problems by the World Health Organisation.

Antenatal and neonatal screening programmes are important in managing haemoglobinopathies and are established in some high-income countries, including UK, USA and parts of the Middle East. However there are very few national screening programmes in lower-income countries in

which these conditions are most prevalent. Local screening programmes are established in areas of many countries and are revealing important information about the microdistribution of these conditions.

In SCD, relatively simple measures, such as vaccination, penicillin prophylaxis, antimalarial measures and treatment of infections are thought to significantly reduce the otherwise very high childhood mortality, although these are not available in many areas. Hydroxyurea is relatively cheap, and has been shown to alter the natural history of SCD, although its role in Africa and India is yet to be established.

The treatment of severe thalassaemia relies largely on the availability of safe blood transfusions and iron chelation, which are available in relatively few Asian countries unless a patient is able to pay. Stem-cell based treatments are becoming increasingly important, and although expensive may be cost-effective in some settings.

Overall haemoglobin disorders cause significant morbidity and mortality across the world. More research is needed to define the prevalence and natural history of these conditions in different countries, and to develop clinical trials and interventions appropriate to low- and middle-income settings.

Session chair: Prof Anuja Premawardena