of its impending demise as a subject worthy of serious study, will not be surprised to learn of yet another book which, like many of its preceding genre, manages nicely to refute the gloomy prognostications.

**Human Hemoglobin Genetics** is a welcome addition to what is already a substantial library given over to this remarkable protein, one of modern biology’s more enduring workhorses. In their preface the authors say they have “endeavored to develop a relatively concise but detailed account of the current state of understanding of the hemoglobin system and the genetic and biological factors that govern its expression”. I think they have succeeded admirably, keeping a nice balance between old and new, so that newcomers in the field are at least aware of the relevant background, and with sufficient detail and breadth to appeal both to students and practitioners. Inevitably, there are deficiencies. Once a topic is broached, where to draw the line can be problematical and I occasionally regretted the restrictions implicit in the title which, while keeping the size down to manageable proportions, put quite a lot of interesting comparative information from other species beyond the book’s scope. A few of the sections, particularly those concerned with the haemoglobinopathies, are really too short to do their subjects justice. But these are minor quibbles that do not seriously mar what is a comprehensive and up to date survey of a very extensive field. There is also a very good bibliography and a useful appendix giving a sort of biological CV on each of the known human globin mutations.

Although not a specialist monograph, my review copy disappeared from my office often enough to suggest that the authors have perhaps been too modest in their expectations of the book’s appeal. For those venturing into haemoglobin research for the first time, whether their interests lie in oncogenesis or population genetics, there is a great deal of interest in this excellent book. Old hands can also profit from it while smugly awaiting the next obituary.

J B Clegg

**Bone Marrow Transplantation for Treatment of Lysosomal Storage Diseases**

The remarkable progress made in recent years into the biochemical and molecular mechanisms of inherited disease has not been paralleled by developments in treatment. It remains a sad fact that curative therapy does not exist for most genetic disorders. Any sound approach to treatment or cure is to be welcomed. Many attempts have been made over the last 15 years to replace missing or defective enzymes in the metabolic storage diseases. To document the history of these attempts is to compile a catalogue of disappointments. However, the advent of bone marrow transplantation (BMT) has justifiably raised genuine hope that effective therapy may be close at hand.

Thus, it is timely that this slim volume, No 22 (1) in the Birth Defects Original Article Series, should now appear, presenting the proceedings of a symposium held in May 1985, attended by representatives of the centres at the forefront of these techniques. The text contains 14 succinct and eminently readable papers, immaculately reproduced on glossy paper and well illustrated. Subjects covered include the mucopolysaccharidoses (MPS), metachromatic leucodystrophy (MLD), adrenoleucodystrophy (ALD), Gaucher’s disease, Lesch-Nyhan disease, Pompe’s disease, technical aspects of transplantation, and studies in animal models.

The message which emerges is one of very cautious optimism. Biochemical correction, as judged by enzyme activity in white cells or plasma or both, was noted for all of the disorders under scrutiny. In several there was also evidence for clinical improvement as judged by resolution of airway obstruction (MPS I), joint immobility (MPS II and VI), ventricular overload (MPS VI), and peripheral nerve activity (infantile MLD).

Unfortunately, however, there was only limited evidence of significant change in the skeleton or in CNS function. No radiographical improvement in bone was noted for MPS II (nine months post BMT) or MPS VI (40 months post BMT). One child with MPS I was found to have normal CSF glycoaminoglycans levels 18 months after BMT, and evidence for arrest of regression based on developmental assessment was presented for five MPS I patients by the Westminster group. The mental state of the five year old child with infantile MLD was much better one year post BMT than that of his untreated sib at the same age, although serial CT scans revealed deterioration. Encouraging progress was also noted in the child with late infantile MLD. Sadly, neurological deterioration continued in the boy with ALD who died 20 weeks after transplantation, and there was no change in neurological status or behaviour in the patient with Lesch-Nyhan syndrome. Nor was any beneficial effect demonstrated in the infant with Pompe’s disease who died 20 weeks after transplant.

There is a clear indication in these reports that BMT has something to offer in the treatment of
lysosomal storage diseases, particularly those lacking CNS involvement. There has been no evidence for neuronal uptake following BMT in animals with lysosomal storage disorders and the evidence in humans is weak. The pioneers of these innovative techniques are to be congratulated. The papers in this text illustrate their awareness of the need for critical, objective, long term evaluation of the patients under their care although fuller data concerning morbidity and risks of mortality would be welcomed.

This book brings together a valuable collection of papers on a very important subject. Its successor providing details of longer follow up is eagerly awaited. Everyone involved in the care of these children will find this volume fascinating.

I D Young

Spina Bifida in South Wales. Can it be Prevented?

The Mainwaring-Hughes Award, donated by the late Mr W T Mainwaring-Hughes, sometime Mayor of the County Borough of Swansea, is used by the University College of Swansea to commission people resident in Swansea or its locality to write pamphlets dealing with contemporary topics in Wales. As one of the regions in the United Kingdom with the highest prevalence of spina bifida and its associated conditions (neural tube defects), these abnormalities are a major contemporary problem in Wales. In the 1960s, one in every 120 babies born had a neural tube defect. Nansi James, who has been assisting Professor K M Laurence of the University of Wales College of Medicine, was invited to write an essay on the research work in South Wales into the cause and prevention of spina bifida.

The essay has nine short sections dealing with topics such as the nature of spina bifida, its effects on both the child and the parents, and the prevention of spina bifida in those who have already had an affected child and also in the population as a whole by screening antenatal patients with serum alpha fetoprotein. To my mind, the most interesting section is the last one-third of the essay which adumbrates the research leading to primary prevention in mothers 'at risk' using dietary management, multivitamin preparations, and folic acid. This section also considers the economic aspects of prevention and looks forward to future strategies for prevention if the Medical Research Council's clinical trial succeeds in showing that folic acid or multivitamins or both can effectively prevent the recurrence of spina bifida and related abnormalities.

The author obviously had difficulty in knowing to whom to aim the pamphlet. From the nature of the background of the award, clearly the object of the essay was the general public. However, the detailed tables found in appendix IV, outlining the various outcomes of the individual studies, are aimed at the scientific audience. In addition, it is difficult to see the purpose, either for the layman or the scientist, of reproducing verbatim the 1720 and 1744 publications from The Philosophical Transactions of the Royal Society describing spina bifida.

It may be mean to cavil at this well meaning effort, but, all the same, it would be negligent for the reviewer not to point out some flaws. The essay as a whole is less evaluative of published reports than one would like, the writer usually choosing to cite relevant works without commentary. Indeed, little recognition is given to other groups, particularly in the United Kingdom, involved in the prevention of spina bifida by periconceptional vitamin supplementation. Flaws in the design and analysis of the South Wales studies are glossed over. However, one cannot but admire the writer's enormous zeal. It is a pamphlet which can be thoroughly recommended to the general public, but not to the specialist in the subject.

Norman C Nevin