of intrauterine and postnatal growth retardation. The Russell–Silver syndrome, and also cases of intrauterine growth retardation without asymmetry, may well be related to constitutional or mosaic uniparental disomy. In mice there are at least six segments of chromosomes which exhibit phenotypic differences depending on whether there is maternal duplication with paternal deficiency or paternal duplication with maternal deficiency. Extrapolating from the man/mouse homologous map one can predict that chromosomes 2p, 5q, 6p and q, 7p and q, 9q, 11p and q, 16p and q, 19q, 20q, 21q, and 22q in humans might show phenotypic differences when there was uniparental disomy for those segments. Again, judging from the mouse and from the human example of cystic fibrosis, one would anticipate growth and behaviour abnormalities but not true malformations in these situations (assuming that a gene that in the homozygous state could produce a syndrome with congenital anomalies was not carried on the particular chromosome). Since both chromosome and DNA markers are available it seems worthwhile to pursue the possibility that patients with Russell–Silver syndrome and other conditions with severe intrauterine growth retardation (where specific congenital anomalies are not present) be evaluated for the possibility of uniparental disomy as the explanation for the intrauterine growth retardation.

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A video presentation ‘Talking about Tay-Sachs’

“It’s not in my family”, or perhaps an admission that you have not heard of the condition before, may be the typical response from someone when asked what they know about Tay–Sachs disease. However, this is no protection against Tay–Sachs occurring in your family.

A 23 minute video presentation about Tay–Sachs and carrier testing is available on loan for a four week period. Families who have personally experienced Tay–Sachs talk frankly about the condition and how it has affected them. Though these parents have suffered the loss of a young child, there is through screening a message of great hope for the future for those watching the programme. Medical and community leaders discuss aspects of counselling and testing which can prevent this family tragedy. Emphasis is placed on the benefits of people knowing their result; before marriage for some, but certainly before starting a family. That carriers are completely healthy and are only at risk of having an affected child if both are carriers is highlighted.

The majority of babies born with Tay–Sachs are born into families with no previous history of the condition. Among the Jewish community carriers are found at the rate of one person in 25. It is for each subject to decide when they would like to be tested, either as a younger single person or when a marriage is planned. We would be pleased to arrange talks with discussion for groups in the UK, show the video, and to offer testing at a later date. Community testing sessions have been particularly well received by younger people.

Details about forthcoming testing sessions are available. If you would like to have a copy of this video, or further information, please contact Zahavah Heckscher or Debbie Seedburgh, Programme Coordinators, Tay–Sachs Carrier Testing Centre.

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This volume was published as the companion to volume 62 in this series and comprises the second part of the proceedings of the 4th Congress of the International Retinitis Pigmentosa Association. It is devoted to issues of direct interest to patients with retinitis pigmentosa (RP) and this is apparent from chapter headings which include: researchers help patients, technical aids, therapy, genetics, living with RP, the RP societies. The majority of contributions are from German authors (the host country for the Congress was Germany) and an attempt has been made to deal with complex issues in simple language. The text is clearly presented and fairly comprehensive in the treatment of the topics which have been chosen for discussion. This book is directed primarily to a lay readership of patients with retinitis pigmentosa and may therefore have a limited appeal to a medically qualified audience.

MARCELLE JAY
A video presentation `Talking about Tay-Sachs'

Ian Ellis

*J Med Genet* 1990 27: 142
doi: 10.1136/jmg.27.2.142

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