

# Celebrating 15 Years of the Gauchers Association

*On the evening Saturday January 20 2007 the Gauchers Association's 15th Anniversary, 138 guests celebrated at a gala Dinner held at the Wembley Plaza Hotel during the week end conference.*

Association Members with family and friends, doctors, nurses and representatives from the Industry attended the Association 15th anniversary gala dinner. Jeremy Manuel OBE, Chairman of the Association welcomed guests.

Keynote speaker Sir Michael Rawlins, Chairman of the National Institute for



*A String Quartet from the Royal College of Musicians entertain the guests during the gala dinner*

Health and Clinical Excellence (NICE) gave the after dinner address and finished his speech by congratulating the Association on what it had accomplished in its 15 years (a full report on Sir Michael's

speech can be found on page 4 of this newsletter). Unfortunately Anne Begg MP who was also due to speak broke her foot on the way to the conference so was unable to attend. Prof Cox however praised the association's work and proposed a toast to the future of the association.

During the day on Saturday 20 January, 65 delegates attended a neuronopathic patient day and listened to presentations on auditory processing; causes and pathophysiology of nGD; results from the recent Type III trial with Zavesca; the blood brain barrier; personal stories; neurological features and complications of nGD and the future of nGD patients. Families included patients from Denmark; Germany and Bulgaria.

On Sunday 21 January the main conference took place 'Studies, Treatment and Costs – Preparing for the Future'. Prof Ari Zimran from the Shaare



*Guests at the Gala Dinner enjoy the evening*

Zedek Medical Centre in Jerusalem gave the opening presentation. Prof Timothy Cox, Dr Patrick Deegan, and Dr Atul Mehta from the UK national Gaucher Centres all gave presentations. Other speakers included Hanna Hyry of Cambridge University, Dr Edmund Jessop from the National Specialised Commissioning Advisory Group (NSCAG) and personal stories from Dan Brown a patient with Type 1 Gaucher disease and Jo Bardoe the mother of Mia Bardoe who has Type III Gaucher disease.

A full report of all the conference presentations can be found in the Gauchers Association 'Conference Supplement' included with this newsletter and can be viewed on the CD attached. In addition you can view the conference in full on the GOLD website at: [www.Goldinfo.org](http://www.Goldinfo.org). Go to the 'Education & Information' section and follow the instructions.



*Families listening to the presentation on neuronopathic Gaucher Disease*



*Young Gaucher patients enjoy the children's activity programme*

# Gala Dinner Speaker Sir Michael Rawlins

*A Gala Dinner was held on Saturday 20 January 2007 at the Wembley Plaza Hotel to celebrate the 15th Anniversary of the Gauchers Association. The guest of honour and keynote speaker at the Dinner was Prof Sir Michael Rawlins, who has been Chairman of the National Institute for Health & Clinical Excellence (NICE) since its formation in 1999, Sir Michael is also the Chairman of the Advisory Council on the Misuse of Drugs and an Honorary Professor at the London School of Hygiene and Tropical Medicine, University of London, and Emeritus Professor at the University of Newcastle upon Tyne.*

Gauchers Association Chairman Jeremy Manuel OBE, in welcoming Sir Michael thanked him for agreeing to be with the Association on their special anniversary and for his support and encouragement during the NICE appraisal into ultra orphan diseases ensuring that the patients view was represented at all times.

Below is the text of Sir Michael's speech to the diners;

'It is both a pleasure and a great privilege to be invited to join you on this special occasion.

'Although I have been practicing medicine for 40 years, I have never looked after someone with Gaucher disease. The reason is because it is very rare. It is described in small print in most medical text books. But one of my old teachers once said "if you have a disease that is small print in the text books it is large print to you" and this is my point. Just because you have a rare disease you are no less worthy of high quality care by doctors, nurses and the NHS than anyone else. And it is a point that the Gauchers Association has made over the years forcefully and effectively.

'Despite my own lack of clinical experience of Gaucher disease I have played some part in its treatment. In the early 1990's, I was Chairman of the Committee on Safety of Medicines (the body responsible for advising on which new drugs should be put on the market and whether existing drugs should stay on the market). During my Chairmanship an application was made to licence Ceredase as a treatment for Gaucher

disease. I remember it vividly for three reasons:

- It was a novel and ingenious approach to treating the condition.
- It appeared to be remarkably effective.
- Only a very few people have been treated with it and far fewer than we would have normally expected. But we recognized that the rarity of the condition made it impossible to expect large numbers.

'One particular concern, though, was that Ceredase was manufactured from human placentas. It needed 20,000 placentas to treat one patient for a year. Our worry was that viral infections might be transmitted to patients. We recognised that the benefits to the patients seemed to outweigh these theoretical risks, so we licensed it. In the event our worries were groundless as no viral infections have ever been passed on. And now the new recombinant product, Cerezyme, has replaced Ceredase which rules out the possibility of viral contamination. Ceredase and now Cerezyme have provided very real benefits to patients.

That might have been the end of my involvement with Gaucher disease and enzyme replacement therapy but, two years ago, NICE were asked how it might approach the evaluation of drugs generally for the treatment of very rare diseases. As part of the investigation we decided to use Gaucher disease and its treatment with Cerezyme as an exemplar. We chose Cerezyme because it



*Sir Michael Rawlins*

had already been used in Britain for 10 years and studying it now would be non-threatening to its continued availability under the NHS. In other words, whatever conclusion we might draw, no one was going to withdraw its use on the basis of our findings.

'Although still unpublished by the Government, we believed (and still believe) that expensive treatments for very rare diseases should be made available under the NHS. This conclusion would I believe safe guard the position of people with very rare disease and, as importantly, offer encouragement to scientists and pharmaceutical companies to continue to develop new treatments. In conducting our enquiry we had enormous help from physicians like Prof Timothy Cox, Genzyme Pharmaceuticals who manufacture Cerezyme, and of course the Gauchers Association.

'Your Association has done an enormous amount for its members. It provides massive support for patients and their families; it has been extremely affective in ensuring the views of patients are heard – and acted on – at the highest levels of the NHS and Government; and it played a critical role in helping us at NICE in developing our approach to the provision of treatment for very rare diseases generally.

'So in paying tribute to the Association, and congratulating you all on what you have accomplished, I ask you to raise your glasses to the 'Gauchers Association'.