

## **2009 AGSD(UK) Conference**

### **Type I and III Workshop**

The morning session was presented by Philip Maes, President of the French Association for GSD (AFG) who covered two areas ; firstly, activities in France concerning GSD and secondly, coordination of GSD activities across Europe.

### **Activities in France concerning GSD**

#### The Association (AFG)

Their website can be found at <http://www.glycogenoses.org>

AFG has 150 patients / 200 members and provides the following;

- information help lines
- information publications
- financing of databases
- limited financial assistance
- contact with medical council
- manifestations (Rallies)
- development of partnerships
- gifts
- influence medical consulting priorities

The medical council (equivalent to AGSDUK's scientific advisory board) now has 8 active members. Membership, activity on their forum and number of website visits have all increased. Their next annual conference / meeting is on Oct 24th / 25th 2009 near Lyon, delegates are welcome from all countries., Dr Weinstein, from the Florida, will be attending .

The IM (Institut de Myology) organises Pompe conferences annually.

#### Medical care

There is a national plan for rare diseases in place. They have Reference and Competence centres which groups GSD medical specialists together. For example, there is a day dedicated to GSD consultation in Lyon. Also the Reference centers have an obligation to help concerned associations like AFG via their medical council. There is a lack of adult specialists and the transition from paediatric to adult is not straight forward. They currently have problems getting Vitaflo's starch approved as a medication.

#### Research

There is research taking place on Gene therapy on mice for GSD1a at INSERM in Lyon and Nantes.

AFG has given 50,000 euros for research to INSERM.

Mice with GSD I cannot be produced naturally so the enzyme has to be blocked in the liver . Mice mature quickly and live for less than an a year so they can be studied throughout their lifespan.

There are databases, now maintained at INSERM. The Pompe database is fully operational and the work has started (using english terminology) to build one for GSDIII.

### **Coordination of GSD activities across Europe**

AFG would like to develop cooperation between AGSD European country associations in order to align respective association objectives. There are GSD associations in UK, Germany, France, Spain and Italy. The International Pompe Association (IPA) is a model for

international working. This would enable smaller countries to have associations with wider reach span e.g. Belgium or Netherlands.

This could be achieved by communicating good practices in Europe, and stimulating international cooperation, producing lists of research projects, creating opportunity for medical staff to leverage internationally, preparing ourselves to represent GSD issues on a European level, share activities when appropriate.

'bad genes don't stop for language barriers'

Activities so far for coordination of GSD activities across Europe include international delegations at country annual conferences, raising of international interest for GSDIII efforts by AFG ex. President Anne Hugon, and financial help to different foundations.

Feedback received so far;

- Germany in favor. Would like to leverage ( Influence) medical council
- Italy: expressed desire to organise European GSD conference, idea favored by IPA
- Spain: wants to leverage (gather) experience from mature associations.

Plans for 2010;

- Collect and publish good practices
- Start a catalogue of research activities in Europe
- Plan for general assembly / committee of representatives from GSD associations
- Organise European GSD conference

Future Ideas ;

- Assemble a Medical Council / Scientific Advisory Board made up of Medical Council / Scientific Advisory Board members from different associations
- Have a separate Medical Council / Scientific Advisory Board at European level
- A country association initially hosts a European GSD conference

-----  
In the afternoon, we heard from the following;

Andrew O'Toole on living with GSD I in his forties. Andrew lives in New Jersey in the USA and is a landscape architect for the New York Parks department. Andrew sees Dr. Weinstein at Duke University in Florida, he takes cornstarch throughout the day and twice at night. Over the last few years he has been on a diet, avoiding fructose, sucrose and lactose but allowing dextrose and glucose. Since starting this diet his liver has reduced significantly in size. How much this reduction could be contributed to the diet was unclear, Dr Phil Lee advised that there isn't evidence to prove this across multiple patients. Andrew takes multi vitamin supplements to make up for the vitamins he misses out on by avoiding the foods containing fructose, sucrose and lactose.

Representatives from Vitaflo told us that they are still in the process of commercially producing glycosade. Several of the families present were using Glycosade and some had had very positive results.although there are several problems still to be overcome

Prof. Smit joined us from Holland, and told us about the databases his team are building for the different types of GSD. He hopes to have 300 patients for the Type III study which is being undertaken at the moment. Patient groups come from Holland, UK, USA and Israel. It is very labour intensive work and not efficient for groups with only a few patents.

On occasions the researcher has gone into a hospital to collect data from the records and this can take several days.

Problems of obtaining Type III data can occur when patients with the disorder stop attending checkups.

Professor Smit hopes to have the study ready for our conference next year.

It was a real pleasure to have Dr. Philip Lee attend the workshop, following his illness. As he's not currently practising medicine, he was not presenting but contributed significantly to the discussions and answered questions from the group on a wide range of subjects such as different views between the UK and the USA on diets (avoiding sugars), quality of life versus rigid management, modified cornstarch, bone density and general trends

Items of interest which occurred during the workshop

1. Eat something before you take glycosade or cornflour.
2. Glycosade has a positive impact on insulin levels.
3. Patients in the USA spend 3 days in hospital when they go for an appointment
4. Beatrix Hospital in Holland has developed a continuous blood glucose monitoring system.
5. Cornstarch versus nasa gastric tube very much depends on the individual patient
6. Swine flu injections should not affect a GSD patient any more than a normal person
7. Indigestion tablets can be taken with a little milk.
8. In Type Ia the muscles can be affected so it is very important to take exercise.
9. In Type I adenomas that are bigger than 5cm or are growing quickly should be closely monitored.
10. Adenomas can reoccur after resection of the liver.
11. Professor Smit found that adenomas did not grow during pregnancy in 15 of his patients but it is not advisable to have an adenoma greater than 5cm when pregnant,
12. Both Professor Smit and Dr Lee agreed that, strangely, those patients who tried to follow the full diet (as stipulated in USA) spasmodically fared worse than those on the full diet and those who did not diet at all.

Finally Professor Smit told us that doctors are getting together to discuss Guide Lines for future reference.

We also need to have an inventory of all the research being done in Europe for GSD, or even better in the whole world.