



## *Rare Disease Day*

The Genetic & Rare Disorders Organisation (GRDO) marked International Rare Disease Day 2011 – Rare but Equal by hosting a patient focused discussion, chaired by Avril Daly, Chair of GRDO, at the Royal College of Physicians on Monday evening February 28. Those present included people with rare conditions, patient advocates, scientists and clinicians. Ms Daly described the Play Decide format which is part of the POLKA Project devised by EURORDIS – the European Organisation for Rare Diseases in conjunction with the Centre for Global Health at Trinity College Dublin as a tool to empower patients and their representatives to become better informed advocates. Play Decide allows a group of people to learn more about an issue through interactive exercises and then decide what their position would be. Four positions are offered as possible outcomes but a group is free to create their own position if they do not form a consensus on those listed. The group then agree on their particular position.

Topics chosen for discussion included:

- Genetic Testing and Genetic Counselling
- Orphan Drugs
- Pre-implantation and Genetic Diagnosis

## **Presentations**

### **Genetic Testing and Genetic Counselling**

Professor Andrew Green Director of the National Centre for Medical Genetics at Crumlin Children's hospital discussed Genetic Testing and Counselling. He gave an overview of the process and mentioned that people receiving results should be fully informed and given sufficient time to understand the implications of their result. It is a breach of the Data Protection Act for GPs to give Genetic Test results without the person receiving the result fully understanding the implications. There are currently 4.7 genetic counsellors in post in Ireland compared with a recommended 32 (Royal College of Physicians UK).

### **Orphan Drugs**

Dr Colin O'Reilly is a practicing hydro geologist and environmental consultant. As a patient with Fabry Disease, he gave a presentation which outlined the definition of Orphan Drugs and highlighted some of the primary issues associated with them.

Orphan drugs are medicines and pharmaceutical agents that are specifically developed to treat rare disorders and diseases. The medical conditions that are treated with the use of orphan drugs are known as orphan diseases. Orphan diseases are highly uncommon and are often genetic. Dr O'Reilly identified the various patient status that exist in relation to orphan drugs as follows;

1. Non-patient
2. Undiagnosed patient
3. Diagnosed + no known drug
4. Diagnosed + drug under clinical trials
5. Diagnosed + existing orphan drug being administered
6. Diagnosed + existing orphan drug not being administered

After describing each status, Dr O'Reilly went on to analyse how the origins of orphan drugs (one or two people working in a lab to cure a specific disease) has now evolved into the modern system we have today which involves multi stakeholders who each have an interest in the area. These stakeholders include the pharma and biotech industries, scientific and academic research teams, medical teams and patient trials, state involvement and of course the patients and families themselves.

The concluding points in Dr O'Reilly's presentation were that:

It is important that we understand the motives of the stakeholders in this movement when engaging in the processes involved for access to orphan drugs.

We must address the fears of a delay in producing and implementing the National Strategy for Rare Diseases – acknowledging what the consequences to further delay might mean to patients. There must be constructive cooperation of all parties at any meetings of the policy working group that is to be established.

Finally, Dr O'Reilly stated that each person in the room was here for different reasons; personal needs, contributions, to learn of progress and to be part of the process.

### **Pre-implantation and Genetic Diagnosis**

Prof. Green then discussed pre-implantation genetic diagnosis. This is an In Vitro Fertilisation procedure for people affected by genetic conditions who wish to have a child free of the condition. There are 7 IVF clinics in Ireland but Pre Implantation Genetic Diagnosis is not available here. It is available in the UK and other European countries with differing regulation standards. As with IVF there is a 20-30% success rate. The process requires a significant time, emotional and financial commitment for the couple.

## Discussions

The discussions were led by Ann Lawlor from the patient support group 22q11, Anna Moran from Fighting Blindness and Karen Pickering from Muscular Dystrophy Ireland. As part of Play/Decide each person takes a different "story card" and reads it to the group so that there is an insight into the various angles around the topic. Information and Issue cards are used to stimulate the discussion.

The outcomes were:

### Genetic Testing and Genetic Counselling

The group favoured a "Fifth" position - one where we have National Regulation with European Leadership / Guidelines which would incorporate a National published Health-Care Policy on Rare Diseases which is regularly updated.

Within this position are the following crucial points

- A National Centre essential to drive policy and guidelines - a central point where anyone can access required information on all aspects of dealing with Rare Diseases
- The development of a post-diagnosis clinical health-care pathways.
- The development of a Public Health Awareness Campaign on rare conditions targeting GP's and Public Health Nurses.
- Genetic Counsellors to be certified - but diagnosis does not always have to come from a Genetic Counsellor, the patient needs to receive results from a skilled health-care professional. A better 'definition' of genetic counselling is sought - psychological counselling can be a greater need.

Cognisance should be taken of the impact on family of diagnosis and guidelines developed on revealing genetic information. Diagnosis should be seen more as a 'process' with practical and human implications. Special attention should be paid to the patient/parent/professional relationship and the importance and need to work closely with RD support groups.

Cognisance should be taken of the barriers and significant cost (emotional/health/financial) benefits of early diagnosis.

### Orphan Drugs - Is there an upper limit on what should be spent on a single patient? The case of orphan drugs

The discussion group consisted of 10 people representing a variety of positions – mostly patients or family members as well as some patient organisation

representatives and medical personnel. There was also academic research representation.

The following positions were discussed:

- Since resources are limited, there should be a limit and only cost effective drugs that benefit the greater number of patients should be reimbursed. For eg If a drug purchaser has to choose between treating 10 patients with drug A or 1 patient with drug B for a rare diseases, then the choice should always be to treat 10 people with drug A.
- Only cost effective drugs should be reimbursed, but the approach for orphan drugs should be different than for common diseases. If there is any doubt about the cost effectiveness of an orphan drug, it should be exceptionally reimbursed in certain cases.
- A position 2 but if there is any doubt about the cost effectiveness of an orphan drug, the benefit of doubt should be given to the patient and the drug should be reimbursed systematically.
- Health is a priority for European citizens. Just as efforts to rescue people after an accident are not restricted, efforts to rescue a patient with a rare disease should not be limited. Cost effectiveness should not be the parameter on which to base reimbursement decisions.

The group felt that ideally they would wish to adopt position 4, however, the reality of the situation demands compromise and therefore position 3 would be most acceptable.

Some points that arose throughout the discussion were:

To be reimbursed by a healthcare system a drug needs to be safe, effective and a cost effective use of public resources. Orphan drugs can not ever be expected to generate sufficient sales to recoup these costs and so should be subsidised and managed centrally. The group emphasised the need to have a transparent and fair system in place to do this.

No price limitation can be put on human life and only through the above method can just decisions be made

The opposition between collective choices and individual preferences was discussed. On the one hand it doesn't seem right to spend many resources for a few people but yet on the other hand it is discriminatory to refuse treatment to an individual who has had no control over their illness. This brought up issues of how public spending is made without question for conditions that may be preventable (smoking related illness, lifestyle related illnesses etc.)

We discussed the "Rule of Rescue" – which is the attempt to help someone in danger, no matter what the cost may be – and how this should apply to the life of a person with a rare disease

There was a discussion on the value of life and how there may be pressure / guilt felt on the part of a patient receiving an expensive, life-preserving drug, to be exceptionally productive in society / life.

### **Pre-implantation Genetic Diagnosis**

There were various scenarios presented to the group discussing Pre-implantation Genetic Diagnosis (PGD) including:

- A family who had a child with spinal muscular atrophy who did not want to risk another child being born with this condition
- A child who was born free of a condition called Fanconi's Anaemia which also meant they were able to provide umbilical cord cells as a treatment for their sibling who already had the condition
- A family who had lost their daughter in a house fire and wanted to access PGD to balance out their family as they already had 4 sons

The group were in agreement that PGD should be available to families who are at risk of having a child affected by genetic conditions and feel that this is the right choice for them. They did not feel comfortable with a policy stating that it can only be used in cases with serious medical consequences, as who decides what conditions are serious enough to meet the criteria? The group did not agree with the use of PGD for choosing gender or personality traits.

When choosing a policy position on PGD, the group favoured policy position 2: "Controls on who can carry out genetic disease testing involving designated professional organisations and a voluntary code of practice. This is managed by an independent body. No licensing".

However, while the group felt that this policy would allow greater consideration of individual needs, it was a concern that self regulation does not always work. They wished to ensure that independent monitoring and evaluation should be carried out in order to ensure the quality of laboratories and outcomes. This should be related to international best practice standards.