

## Rare Disease Day in Wales

Thursday 4th March 2010

A group of one hundred families, clinicians, researchers and politicians in Wales came together to mark Rare Disease Day 2010 with a reception at the National Assembly for Wales on Wednesday 24th February. Rare Disease Day is part of a global initiative to raise awareness of rare diseases and improve and coordinate services for people living with rare conditions and their families. The event was organised by Rare Disease UK and hosted by Helen Mary Jones AM for Llanelli.

Several families from Alpha 1 Awareness UK attended the event including Jeff & Meryl Darkins from Blaenau Gwent and also Angela & Ray Miller from Maesteg.



Photo: Jeff Darkins, Trish Law and Meryl Darkins

Rare Disease Day 2010 was on Sunday the 28th of February and the theme was 'Patients & Researchers: Partners for Life' and raised awareness of the need for research into rare conditions and development of treatment and support for people living with them.

A rare disease is one that affects about one in 2,000 people. There are more than 6,000 different rare conditions. Rare diseases are surprisingly common – 3.5 million in the UK (175,000 people in Wales) - live with a rare condition at some point in their lives.

Rare Diseases are diverse in severity and symptoms presented by patient. People living with these conditions, however, report similar issues around diagnosis and treatment. It can take patients years to achieve a diagnosis, leading to a deterioration of the condition, inappropriate treatment and medication and significant distress for them.



Photo: Angela Miller and Janice Gregory

Rare Disease UK is campaigning for a strategy in Wales and across the UK for integrated service delivery for rare diseases to aid speedy diagnosis and effective treatment and care for people living with rare conditions.

Alastair Kent, Chair of Rare Disease UK, said: "There are tens of thousands of people in Wales and millions of people in the UK living with a rare condition. For many, there is little knowledge on

how to treat and manage their condition and scientific research is lacking. Raising awareness of rare diseases and the need for a co-ordinated approach for the diagnosis, treatment and research of rare diseases is hugely important.”

Rare Disease Facts:

- 75% of rare diseases affect children and 30% of rare disease patients will die before their fifth birthday
- 1 in 17 people will develop a rare condition at some point in their life
- Rare diseases are often chronic, progressive, degenerative, and often life-threatening
- Rare diseases are disabling: the quality of life of patients is often compromised by the lack or loss of autonomy
- Rare diseases are characterised by a broad diversity of disorders and symptoms that vary not only from disease to disease but also from patient to patient suffering from the same disease. Relatively common symptoms can hide underlying rare diseases, leading to misdiagnosis.
- Rare disease patients face common problems:
  - Lack of access to correct diagnosis
  - Delay in diagnosis
  - Lack of quality information on the disease
  - Lack of scientific knowledge of the disease
  - Heavy social consequences for patients
  - Lack of appropriate quality healthcare
  - Inequities and difficulties in access to treatment and care