

L - R: Event chair professor Reinhold Schmidt, Vicki Modell, co-founder of the Jeffrey Modell Foundation (JMF), MEP Miroslav Mikolášik, JMF co-founder Fred Modell, Grigorij Kogan from the European commission



# Deficient diagnosis

MEPs have joined forces with health experts in demanding EU-wide action to raise awareness of a little-known but potentially deadly disease. **Martin Banks** reports

It is said to affect up to one in 250 people. It is often debilitating and sometimes even fatal. But a recent debate in the European parliament heard that relatively little is known in Europe about primary immunodeficiency (PID), a deficiency in the immune system that causes recurring infections.

The lunchtime event, organised by Brussels PA company Interel, in parliament last week was told that because of such poor awareness, Europe lags well behind other parts of the

## Warning signs

Patients, whatever their age, affected by two or more of these warning signs should be tested for PID:

- Four or more new ear infections within 12 months
- Two or more serious sinus infections with one year
- Two or more months on antibiotics with little effect
- Recurrent, deep skin or organ abscesses
- Two or more deep-seated infections including septicaemia

world, such as the US and Asia, in the treatment available for people with PID. The message, therefore, was clear: the EU and member states need to step up to the plate and focus more attention on improving patient care.

According to Slovakian EPP member Miroslav Mikolášik, who opened the debate, the key is early diagnosis and equal access to treatment. The sooner the condition is diagnosed, the better the chances of survival. Member states, he said, also have an important role to play in alleviating patient suffering by including PID in all newborn screening programmes for rare disorders.

Another keynote speaker, Jose Drabwell, spoke eloquently and passionately about how it had taken “years and years” before she was diagnosed with PID. She encouraged member states and MEPs to sign a “call to action” which explains the steps national governments can take “to understand, appropriately diagnose and manage PID”. Drabwell, who chairs PID’s International Patient Organisation, said,

*“Many people with PID die at a young age because the disease is not detected and their healthcare practitioner did not recognise that these recurring infections were caused by a deficient immune system, or PID”*

“The PID community is calling upon member states to take heed of these devastating and under-diagnosed diseases by developing national rare disease plans that include PID and ensure that both new and existing national rare disease plans cover PID.”

The event was jointly organised by the Jeffrey Modell Foundation, founded by Fred and Vicki Modell in memory of their son, Jeffrey, who died at the age of 15 from complications associated with PID. Fred explained how a deep personal tragedy had driven the couple to act to help prevent others suffering in the same way. He said, “Jeffrey was diagnosed when he was just nine-months-old but, fortunately, was able to go on and lead a relatively normal life.” Their son eventually succumbed to the condition, dying from severe pneumonia. His father said, “In a way I suppose he was lucky because it is thought that up to 90 per cent of people with PID remain undiagnosed. They just wallow in sickness without ever knowing what is really wrong with them. Many people with PID die at a young age because the disease is not detected and their healthcare practitioner did not recognise that these recurring infections were caused by a deficient immune system, or PID.”

Professor Reinhold Schmidt, of Hanover Medical School in Germany, who chaired the debate, said that a “simple and relatively inexpensive” blood test called “complete blood count” can identify over 95 per cent of affected patients. “These tests are widely available in Europe,” he said. “It is awareness of PID that is absent and begs improvement.”

Modell stressed that the PID ‘community’ does not seek financial contribution from the EU. “The intended purpose of this event is to solicit the support of the EU in connection with earliest possible diagnosis, appropriate treatment, physician education and public awareness of PID in Europe,” he said.

Participants heard that the number of PID patients varies significantly between member states, from five per million in Germany to 47 per million in Sweden, which, for no apparent reason, has the highest prevalence. Professor Lennart Hammarstrom, of Karolinska University in Huddinge, Sweden said this begs the question whether member states are using different diagnostic criteria. He says that while the EU may not have a responsibility for healthcare, it still has an



“important” role to play in improving and safeguarding public health. “Raising awareness and encouraging health systems to act can make a difference and create a better quality of life for people living with PID,” he said. ★

#### Current scientific estimates of the rate of PID show:

- Around one in 8-10,000 people has a genetic primary immunodeficiency that significantly affects their health
- PID affects at least 10 million people worldwide
- Experts estimate that the actual incidence of some form of PID in the general population is between one in 250 and one in 500
- This compares with a prevalence of one in 700 for type one diabetes and one in 1000 for multiple sclerosis
- While each of the more than 160 identified PID conditions may be rare, taken together, they are more common than childhood leukaemia and lymphoma combined