Directorate-General for Health & Consumers

EU Health Prize For Journalists 2009

National Jury

Chair:

Carsten Lietz

Press attaché
European Commission representation in Germany.
Leiter der Pressestelle und Pressesprecher der Kommissions-Vertretung in Deutschland

Hubertus von Voss

Dr. von Voss is head of INSOPA (Institute for Social Paediatrics – Rehabilitation) at the Centre for Human Genetics and Laboratory Medicine in Munich.
He studied medicine in Freiburg and Munich and paediatrics in Düsseldorf, followed by post-doctoral studies. Spent 10 years as Head Doctor of the Kaiserswerth Church Paediatric Hospital and the "Florence Nightingale" institutes in Düsseldorf.
1990-2008 - Professor for Social Paediatrics at Ludwig–Maximilian University in Munich and Medical Director of the Munich Children's Centre (funded by the Upper Bavaria district authorities). On scientific advisory boards at rehaKIND, Internationale Fördergemeinschaft Kinder- und Jugend-Rehabilitation e.V. Joint founder and president of KINDERNETZWERK e. V. since 1991.

Marlinde Lehmann

Since 2001, Dr. Lehmann has been head of the medical department at Ärzte Zeitung, the exclusive German medical daily newspaper for doctors, focussing on new findings in neurology, psychiatry and gastrointestinal diseases.
Aside from her career as a journalist, Dr. Lehmann has worked for several years in clinical research and has been involved in medical development aid in Guinea, West Africa.

Andreas Mihm

Correspondent for the Frankfurter Allgemeine Zeitung, a major German daily newspaper, in Berlin since 2001, focussing on health, economic and energy policy.
Mr. Mihm started his journalism career in 1988, working for WDR (Western German Broadcasting) and Reuters. In 1990 he moved to Berlin. He reported on the German reunification process and the economic development of Eastern Germany for Reuters as well as for the "Neue Zeit" until 1994. In 1997 Mr. Mihm became head of the Berliner Zeitung's economics desk. Later, he moved on to the Handelsblatt, where he worked as head of the department dealing with issues concerning Berlin and the New Länder until 2001.

Franz Porzsolt

F. Porzsolt, MD, PhD, has been trained in tumour biology (Ontario Cancer Institute,Toronto, 1974/75), internal medicine, haematology and medical oncology (Medical SchoolUniversity of Ulm). He worked as a clinician from 1976 through 1996 and was head of the Cancer Center at the University of Ulm. Since 1986, he has been deputy director of the Department ofHaematology/Medical Oncology at the University of Ulm. Based on the experience as medical oncologist he started to work on methods that would allow comparison between what patients receiving cancer treatment have to „pay" and what they„get back" (in intangible not monetary units). He calls this new field „Clinical Economics".
As quantity and quality of life are the primary endpoints from the patient´s point of view, F. Porzsolt has also gained experience in experimental quality of life (QoL) research and is member of the QoL study group of the European Organization for Research and Treatment of Cancer (EORTC). His second and presently dominant field of research and teaching is Evidence-Based Medicine (EBM). He was trained in EBM at McMaster in Hamilton and in Oxford and has been member of the Centre for Evidence-Based Medicine in Oxford since 1997.

Germany

National winner

Katharina Kluin

Katharina Kluin is working as an editor for the German news magazine Stern for the last two years, covering health and science topics.

She started working as a journalist while studying information management. She completed her professional training at the Henri-Nannen-Schule in Hamburg.

In 2008, she travelled and worked in South America, mainly Brazil. She is 29 years old and lives in Hamburg.

Original article: Waisen der Medizin

English translation: Medicine's orphans

The article presents a widely unknown problem both in journalistic and professional terms in a very sophisticated manner. It refers to one of the issues of the Europe for patients' campaign and highlights the national as well as the European and international dimension. It calls for more research and reassures patients concerned. The article is written in a very comprehensive and intoxicating way and is well investigated.

Authors: Katharina Kluin

National nominee for Germany

Media: Stern, 26/02/2009

Medicine's orphans

They're only a minority. And yet there are a lot of them: over 4 million Germans suffer from a rare disease. They often have to wait years the right diagnosis and there are hardly any treatments. Often, their only source of comfort and hope is self-help groups.

'I'm looking forward to my next hand op', says Hannah, and she really means it. During the next operation, the surgeon will separate her fingers, as they keep on fusing together. He will graft skin on to her palms so she can stretch out her joints again and put thick wires into her fingers to stop the new smooth skin from tightening up again. And she's looking forward to that? 'Oh yes, I really am', says Hannah, with her eyes sparkling. 'And then I'll get the next *Twilight* book.'

Hannah-Lisa Daby from Delmenhorst, 12 years old, was born with *Epidermolysis Bullosa* (EB). The different layers of her skin lack the anchors that should hold them together. The slightest firm contact, a rub or knock, causes blistering and even open wounds. Her throat and oesophagus are also affected. Because of scars and deformation, she can't open her mouth more than 1.5 cm. But the first thing you notice is her hands. Because her thin, tightly-stretched skin undergoes a constant process of forming blisters and healing, she's lost all her nails and her fingers are misshapen and, in parts, fused together.

There is no cure. The disease is so rare that doctors are often at a complete loss when faced with an EB child. So rare that very few scientists show any interest in it and that for pharmaceutical companies, it's hardly worth searching for a remedy. An illness is considered rare if it affects fewer than 10,000 people. That's how a small number of 'rare cases' mounts up: in Germany, over 4 million people suffer from one of 5 000 rare diseases.

Most patients have exactly the same experience: they wait years for a diagnosis, drive hundreds of miles to see a specialist, find too little information on their disease and have to live with the fact that most of the time, research on potential cures simply isn't profitable enough. These illnesses all look very different, whether they cause muscle atrophy, immune-system failure or sudden bone overgrowth.

'Mummy, why's the baby all broken?', Hannah's brother asked when he saw his little sister for the first time. She was a veritable bundle of bandages, with compresses around every single finger, and her feet and knees. A thick drop of blood ran down Hannah's little wrist when she grabbed her rattle for the first time. When she started exploring her nursery on her knees, her mother lay mattresses on the floor as a precaution.

A few months later, she also covered the walls with mats. During the first three years of her life, this four-by-four foam-lined room was the only place where Hannah could move about safely. 'I built a padded world around her', her mother says. During her short outings in the world beyond the cotton microcosm, she had to hold Hannah's hand very loosely; firmly enough to make sure her daughter was walking safely by her side – but gently enough not to tear the skin off her hands if she fell or stopped walking.

Like 80% of rare illnesses, EB is the result of a

genetic disorder which is either inherited or caused by spontaneous mutation. Doctors have established that the two mutated genes responsible for Hannah's destiny do not belong to the more or less 700 known EB genetic variants. 'Brilliant. And where does that leave us?, Hannah asked at the time. 'We now know that you're mutation 701 and 702', Sabine Daby said, and then put the results away in one of the numerous files in the two bookcases filled by her daughter's disease.

When talking about rare illnesses, specialists often use the term 'orphan diseases' – orphan because nobody cares about them. Here and there, a team will do some research on something or other, and the few specialists that do exist are scattered all around the world. Decades can go by before the bits of the puzzle actually build an overall picture than can help patients.

In Germany, about 90 patient groups have united in the 'Achse' - the German Alliance for Chronic Rare Diseases – to raise awareness of their problems. The organisation will be celebrating European Rare Disease Day on 28 February with events throughout Germany. The patron of the organisation is Eva Luise Köhler. Her daughter Ulrike lost her sight to a rare ophthalmic disease. 'What we want to achieve is that people with rare chronic diseases are treated like any other patients', says the German president's wife (see interview, page 80).

Knowing that several million sufferers can be influential definitely helps the 'Achse' and other European self-help groups. This lead the EU to create a Rare Diseases Task Force and a strategy to make developing special medicines more advantageous. The German government also wants to invest 80 million euros by 2020 into research projects that take an interdisciplinary approach to causes, therapies and medical care – and thus view rare diseases as a whole rather than individually.

Treatment is indeed complex in most cases. Whereas Hannah-Lisa Daby's specialists are at the Freiburg University skin clinic, her hand surgeon is in Münster and her pain therapists in Datteln.

Since brushing her teeth too firmly is enough to make her gums bleed and cause blisters, the 12-year-old also has to have her teeth cleaned professionally every three months. A dentist in Delmenhorst took the child back to her mother, who was sitting in the waiting room, without actually having done anything and blurted out she never wanted to see her again. Since then, Sabine Daby has had to take Hannah to a dentist specialising in EB in Herten, a little town in western Westphalia, 200 kilometers away – and one of the only ones in Germany.

Generally speaking, there are very few experts who specialise in care for sufferers of rare diseases. Physiotherapists, nursery-school teachers and teachers too often lack the knowledge to deal with such conditions – how could they if this is the first time they've encountered such a disease in decades of working? For sufferers, this can spell utter horror – for Hannah too, who had to pick out clay from the open skin on her palms after her teacher at kindergarten let her play with it.

Nowadays, Hannah mostly goes out with Einstein, her dog. This specially trained Elo can open doors, which Hannah can't because of her hands, and he is her companion on a road to a more independent lifestyle. A companion who knows he shouldn't pull on the lead, which she only wraps loosely around her forearm.

In the company of Einstein, Hannah can go for walks around town and through the park, and recently, after school on Wednesdays, she has been venturing as far as the newspaper kiosk to buy 'Bravo', her teenage magazine. The two 'Twilight' double posters she got from a previous edition are now hanging above her reading chair and her desk. The dark picture has 'Love kills' written on it. She says she likes it.

Her disease makes her feel lonely. Hannah-Lisa is sometimes one of the only ones in her class not to be invited to birthday parties. Perhaps it makes the other children feel uneasy, perhaps their parents are afraid of being responsible for a sick child. Two or three girls from her class go into town with her from time to time.

But the only friend who was always by her side during her childhood, 'the best friend I can imagine', moved to Australia with her parents two years ago. Since then, Hannah has slipped every single cent of pocket money that's left into her 'Australian box', the piggy bank collecting money for a ticket to the other side of the world.

Susan Wanzenberg (37) has also seen her circle of friends shrink since becoming a rare case. Living in Schwerin (northern Germany), she suffers from systemic Lupus Erythematodes (SLE), a serious rheumatic disease affecting no less than 40,000 patients throughout Germany. A lot of time when by before she found out.

She was in her mid twenties when the disease emerged. She spent two years numbed with fatigue, suffering from joint pains and bouts of fever for months at a time. After numerous wrong diagnoses – ranging from bronchitis to appendicitis and a virus brought back from holiday – she suddenly lost consciousness on a January day in 2000. The emergency doctor she called twice diagnosed a serious case of flu. She should sleep it off, he said.

After she failed to wake up for two days, her parents insisted on taking her to hospital. The doctors there discovered her immune system was life-threateningly hyperactive; it had obviously turned against her own body. At first, they suspected leukaemia. It was only when a specialist from the rheumatology department was called that she was diagnosed with Lupus Erythematodes. At that time, a war was already taking place inside Susan's body, having given her pericarditis, liver failure and bouts of drowsiness. Two more days and the diagnosis would have come too late.

But this one saved her life. Two years of chemotherapy followed. It brought an end to the chaos in her body's defences. Nine years on, she still has to take cortisone to contain the joint inflammation that keeps flaring up and medication to inhibit her immune system. Going to the cinema, to a village fair or having a walk in the Schlosspark shopping centre in Schwerin is out of the question during the winter months, when she could

easily catch a cold. The medication she's on suppresses her natural defence against infections so she's just not sufficiently protected. And she's not just at risk from other people – even sunshine can trigger a dangerous reaction. This is why, even in winter, Susan wears sun-tan cream, and in summer she sits on a north-facing terrace.

Thanks to her medication, Susan is mostly able to cope with everyday life without assistance. She goes on long walks with Carlson, her golden retriever and does the housework in her newly built house in the Mühlenberg area of Schwerin. But because her illness is often unknown to doctors and mostly goes unnoticed in public, her greatest battle is often to make people understand.

Even her gentle routine makes her feel like she used to after two hours of squash. To do any other type of activity, she has to prepare herself by taking a long break. Sometimes the inflammation in her fingers makes them so stiff she loses all feeling in them. All this is what outsiders don't see. They shake their heads at the sight of this strange-looking woman covered up in long clothes in the midst of summer, walking around in a big summer hat not doing much - but leading a nice life at public expense.

Even the rheumatologist who was supposed to assess Ms. Wanzenberg's work capability for the German pension authority didn't understand what he was dealing with. 'We're all tired from time to time, I'm also tired all day long', he said to the bewildered patient. 'You're a big and strong woman, why couldn't you work at least part-time?'. Having been awarded provisional early retirement in 2000, every two to three years Susan must prove she is not able to work. In this case she was only able to reverse the rheumatologist's assessment by providing a costly counter-opinion.

This lack of recognition among doctors is typical. A survey of 6,000 sufferers conducted by the European Organisation for Rare Diseases has shown that one in four of them had to wait even longer than Susan Wanzenberg for a diagnosis - between five and thirty years. And when there was an early diagnosis, in a third of cases the doctors were wrong. In such cases, the patients were treated for a disease they weren't suffering from. In the worst cases – experienced by some 16% of patients questioned - this even led to an unnecessary operation.

And to every third patient, doctors prescribed medication they should never have been given. Like Susan Wanzenberg, many sufferers are regarded as malingerers, or doctors insinuate the cause of their physical pains is psychological. According to this study, 10% of patients with rare diseases end up receiving psychotherapy before getting a proper diagnosis.

Florian Frei* suffers from Von-Hippel Lindau disease (VHL), a rare illness where tumours constantly appear in various organs. He was 26 and in the middle of his studies when the first tumour was removed from his cerebellum. That was 14 years and 24 tumour operations ago - eleven in his brain, five in his eyes and spinal marrow, one in his inner ear, four in his left kidney and three in his right. 'At the moment, another one is growing in the cerebellum, a few in my back, one on an optic nerve, and now there are also two in my pancreas, which is very new', he says, picking up his cup of tea unmoved.

This movement seems as difficult as his breathing when he speaks.

He also spreads his fingers a little too much, lifting his cup to his mouth in a slow, sweeping movement. 'I really have to concentrate', says the Cologne-born man, and every single word he pronounces seems to take as much effort as picking up the mug. The cerebellum operation before last altered his balance, his fine motor functions and his ability to speak and swallow slightly. 'The tumour was gone and I was still alive: from a medical point of view, the operation was a success, I was told. Obviously, I see things differently.' Before he had this operation two years ago, Florian Frei must have been an eloquent man. His eyes want to say much more than his tongue can. And in his case too, there is no cure. 'The only therapy is the scalpel, he says. Research on this disease is difficult because hardly any of the cases bear any similarities to others – and the possibility of gene therapy to cure it is still highly unlikely. In Florian's case too, a spontaneous gene mutation seems to have caused this tumour syndrome. Moreover, not all developments in cancer research can be applied to VHL sufferers. Only kidney tumours, which are typical for VHL, are considered a type of cancer. And whereas other cancers are operated on immediately, doctors tend to look at VHL tumours for months or even years. Most of the time, they only reach for the scalpel when the tumour is so big it hinders organ function or risks spreading.

To make sure it isn't suddenly too late, Florian Frei is pushed into the same tube every few weeks. With computer tomography and other imaging technologies, his doctors watch the unbridled spread of tumours through his body. Every one to two years, an operation is carried out. For him, his partner and family, this is always nerve wrecking. Indeed, nobody knows in what state he will wake up. And he only wakes up to wait for the next tumour to be found.

'When things get critical, he says, that's when we have to start struggling with the inadequacies of the system, which simply cannot cope with such rare diseases.' Every single time he has to have a tumour removed from his head, spinal marrow, kidneys or wherever, he has to justify to his health insurance why treatment can only be carried out by this or that clinic, why travel expenses or costly physiotherapy have to be paid for. 'Every time, I have to explain the specific problems caused by my specific disease – and that not any old doctor can deal with it.'

In all these years of fighting, he's 'developed a thick emotional barrier'. A barrier equipping him for a life with this illness. 'Life goes on, whether I like all this or not', he says. 'Nowadays, the life expectancy of VHL sufferers is actually the same as anybody else's. And that's something I'm not going to question.'