Background paper

of the campaign „Rare diseases - not at all that rare“

initiated by dsai

www.dsai.de

Contents

1. Welcome ▲

Helga Kühn-Mengel MdB
German Federal Government Commissioner for Patients' Affairs

Dear Reader,

The topic of rare diseases is one to which I devote particular focus, for this is an area that receives little attention amongst the wider public. Indeed, even in medical circles, a great deal of information on the subject is lacking. Many of those who suffer from rare diseases find themselves in a difficult situation both within the health care system and within society, for the causes of their suffering are recognised much too late or are not diagnosed at all.

Thus, as the Federal Government's Commissioner for Patients' Affairs, I welcome the campaign of the German Self-Help Organisation for Congenital Immunodeficiencies e.V.: "Rare Diseases – Not at all that rare: Early diagnosis saves lives and reduces treatment costs". The campaign seeks to educate the public and has drawn up a list of concrete measures to improve the situation of sufferers and to relieve the long-term burden upon the health care system. It argues in particular for early diagnosis, appropriate treatment and comprehensive, area-wide care for sufferers.

Every patient rightfully expects that all diagnostic and treatment possibilities will be exhausted in order to offer them sound medical care, irrespective of their level of income or education. The health care reforms of recent years have set a course towards better quality and transparency in health care. In order to integrate and strengthen patients' interests within the health system, policy makers are reliant upon the contributions of support groups and patients' organisations. And precisely with the example of rare diseases, it becomes clear that politics, science, research, practitioners, industry and those affected must all work together intensively to ensure the best level of medical care.

It is thus that I wish the campaign and all those affected by rare diseases every success in bringing about their goals.

2. Foreword ▲

Professor Dr. med. Reinhold E. Schmidt
Director of the Department of Clinical Immunology at the Centre for Internal Medicine of the Medizinische Hochschule, Hannover; President of the German Society for Immunology (DGfI) and member of the board of the European Federation of Immunological Societies (EFIS).

Dear Reader,

The German Self-Help Organisation for Congenital Immunodeficiencies has initiated the campaign “Rare Diseases – Not at all that rare: Early diagnosis saves lives and reduces treatment costs” to educate the public about primary immunodeficiencies (PI) in particular.

In Europe, the number of sufferers stands at 1.5 million, and 95 per cent of patients with PI can be diagnosed with a relatively simple and inexpensive bloodtest. In Germany, the number should be assumed to stand at 100 000 patients, of whom only a fraction are diagnosed and adequately treated. Within the EU there are large differences in treatment and no uniform coverage with medications. Specialist knowledge and information are often simply lacking within medical circles.

I expressly support this campaign because primary immunodeficiencies receive hardly any attention within the EU health care system. If the diagnosis rate of primary immunodeficiencies does not considerably improve, then the long-term treatment costs will increase with misdiagnosis. An early diagnosis is clearly more cost-effective. The goal of the campaign is to improve fundamentally the quality of life of sufferers. With our demands to implement three concrete measures we would like to send a signal to the Federal Government and make a contribution towards the increased efficiency of the health care system in Germany.

3. Introduction ▲

Gabriele Gründl
Chairwoman, German Self-Help Organisation for Congenital Immunodeficiency e.V. (dsai)

People with a rare illness exist on the margins of the German health care system. This is the case, even though rare diseases are in no way a rarity! With the campaign **“Rare Diseases – Not at all that rare: Early diagnosis saves lives and reduces treatment costs”**, sufferers have joined together under the auspices of the German Self-Help Organisation for Congenital Immunodeficiencies e.V (DSAI e.V.) to inform and educate policy makers and the public about rare diseases, and about primary immunodeficiencies in particular. Our campaign is supported by the Alliance for Chronic Rare Diseases (ACHSE e.V.), the German Society for Immunology (DGfI), the European Federation of Immunological Societies (EFIS), the European Organisation for Rare Diseases (EURORDIS) and the International Patient Organisation for Patients with Primary Immunodeficiencies (IPOPI).

Awareness within German politics and amongst the public about rare diseases and primary immunodeficiencies is too low. Rare diseases have up to this point figured only marginally on the health care agenda and in discussions concerning health care reform. Even within the medical

establishment, specialist knowledge and important education programs about rare diseases are often lacking.
Rare diseases are often life-threatening and can cause chronic disability. In spite of this, they are only seldom diagnosed and patients are left to suffer on their own.

It can be assumed that a total of 30 million people within the EU have a rare disease. Approximately 4 million of these people live in Germany. Out of these 4 million sufferers, calculations suggest approx. 100 000 have a congenital primary immunodeficiency condition. Yet, based on our information, only approximately 700 patients have been diagnosed as having this primary immunodeficiency.

On an EU level, on the other hand, the topic of rare diseases is given more recognition than it receives within German politics. The health care strategy of the European Union is already pointing in the right direction: the Public Health Action Program of the EU for the years 2003 to 2008 contains a chapter on rare diseases. This forms a good foundation upon which the EU can promote research programs, conferences and concrete projects, such as, for example, the development of a European database for rare diseases and the EU Commission’s “Rare Diseases Task Force”.

In order to assert a stronger profile for rare diseases within politics and society in Germany, we have come up with the **Berlin Signal**. The **Berlin Signal** contains the goals of our campaign and a catalogue of concrete measures to improve the situation of sufferers in Germany. The **Berlin Signal** will be sent to the relevant decision makers in politics and health care and should serve as the basis for discussions in the political arena.

Gabriele Gründl

4. What are rare diseases? ▲

4.1 No rarity: Rare diseases ▲

Rare diseases form a heterogeneous group of very different conditions. It is estimated that there are between 5000 and 8000 rare diseases. Examples of these include cystic fibrosis, hemophilia and primary immunodeficiency conditions. Most types of cancer, including all childhood cancers, fall under the category of rare diseases.

According to the EU definition, a disease is considered “rare” if less than one out of every 2000 people suffers from it. [1] In other words, for every one million citizens there are as many as 500

[1] EC Regulation on Orphan Medicinal Products.

patients with a rare disease. Whereas the figure of 1 in 2000 might seem small, it can mean that with a total population of 459 million citizens in the EU, up to 230 000 could suffer from a rare disease.[2]

The majority of rare diseases assume a severe, chronic and sometimes life-threatening course. 80% of rare diseases are genetic and particularly affect children and adolescents. Many of these illnesses are incurable and most seriously impact upon the quality of life of patients and those around them.

The rarer a disease is, the harder it is to diagnose and the worse the quality of the treatment is which it receives. A frequent cause is the information deficit currently existing among basic care providers about available specialist treatment facilities. Often, even in specialist centers, the level of knowledge of extremely rare diseases is insufficient. In addition to this, and all too often, effective treatment cannot be offered. This is also due to the fact that, for these conditions, research is more rarely produced.

Congenital immunodeficiencies are a classic example of a group of rare diseases.

4.2 A classic example: Primary immunodeficiencies ▲

Patients with a primary immunodeficiency are rendered defenceless to infections because their body’s immune system is weakened. 128 different immunodeficiencies have been identified thus far, whose effects can often be life-threatening when incorrectly diagnosed and treated.

Each of the various primary immunodeficiencies needs specific treatment. In many cases, treatment consists of regular, life-long immunoglobulin replacement therapy and targeted treatment of specific pathogens with antibiotics where there is an acute infection. An incorrect course of treatment or diagnosis can lead in the majority of cases to chronic infections, resistance to antibiotics and life-threatening organ damage.

In spite of the often conspicuous, life-threatening clinical symptoms, the primary deficiency is often not recognized.

4.3 The figures at a glance ▲

Rare diseases:

- Between 5000 and 8000 different rare diseases have been identified
- 80% of rare diseases can be attributed to genetic causes.

[2] “Rare Diseases : Understanding this Public Health Priority”, Eurordis, November 2005, p. 3, http://www.eurordis.org.

- In 50% of cases, symptoms of the disease begin in childhood.
- Most types of cancer, particularly childhood cancers, are rare diseases.
- In all, there are 30 million sufferers in the 25 EU States.[3]
- 6–8% of the total population of the EU are affected.
- In Germany, around 4 million people are affected.

Diagnosis:

- The number of diagnosed patients in Germany and Europe is not known.
- European studies suggest that the proportion of error upon initial diagnosis of rare diseases is approximately 40%.[4]
- In Germany, the proportion of misdiagnoses according to this study is as high as 50%.

“Diagnosis is determined late or not at all.”[5]
Anne Kreiling, Chairwoman of the Alliance for Chronic Rare Diseases e.V. (ACHSE)

Primary immunodeficiencies:

- In contrast to secondary immunodeficiencies such as AIDS (Acquired Immunodeficiency Syndrome), PI’s are caused by innate or genetic defects in the immune system.
- 128 types of immunodeficiencies have been identified. Among the best known primary immunodeficiencies is severe combined immunodeficiency (“bubble boy disease”).
- In the EU, about 1.5 million patients fit the clinical picture of having a primary immunodeficiency.
- In Germany, the number is approximately 100 000 patients.[6]

Diagnosis:

- So far, only about 700 of the 100 000 patients in Germany have been diagnosed.[7]
- Germany is currently in second-last place in terms of the rate of diagnosis of primary immunodeficiencies in a Europe-wide comparison.
- 95% of primary immunodeficiencies could be diagnosed by way of a simple blood test.[8]

“One of the biggest misunderstandings is that immunodeficiencies are so rare as to be negligible. Even if one doesn’t count a selective IgA deficiency with a prevalence of 1:500, primary immunodeficiencies in the order of approx. 1:2000 can be expected. This is an estimate, and precise figures are not available. But this would mean that an average of one patient with PI is treated in every doctor’s practice.”
Prof. Dr. Volker Wahn, Campus Virchow-Klinikum Berlin, Pediatric Hospital specializing in Pneumology and Immunology

[3] WHO, “Priority Medicines for Europe and the World Project ‘A Public Health Approach to Innovation’ ”, Ch. 7.5 “Orphan Diseases”, 7. October 2004, p. 5, http://mednet3.who.int/prioritymeds/report/index.htm.
[4] Eurordis Study “EurordisCare 2: Survey on the delay of diagnosis for 8 rare diseases in Europe”, 2004.
[5] Patientenbrief, Ihr gesundheitspolitischer Fakten- & News-Letter, GSK, May 2006, p. 7.
[6] Prof. Reinhold E. Schmidt, European Primary Immunodeficiencies Consensus Conference, 19.-20. Juni 2006.
[7] Estimates of the German Self-Help Organisation for Congenital Immunodeficiencies, http://www.dsai.de.
[8] Prof. Dr. Reinhold E. Schmidt, Foreword to this Background Paper.

Conclusion: Although each individual illness is “rare”, there are millions of patients with rare diseases. In addition to this, the still greater number of family members and friends who are indirectly affected by the illness should not be forgotten.

5. Living with a rare disease

5.1 On the margins of health care: Lack of awareness

Rare diseases, in particular primary immunodeficiencies, do not feature in political and public discussion in German. The topic of rare diseases is given little prominence on the health care policy agenda or within the debate surrounding health care reform.

Rare diseases are dealt with in small circles at medical conferences and in support groups. In Germany, there is currently neither a state-sponsored central information pool, nor a public information campaign about rare diseases.

Thus there is still a need for comprehensive work nationwide to educate and inform policy makers and the general public about rare diseases.

> *“The significance of rare diseases is underestimated.”*
> Prof. Karl W. Lauterbach MdB, Institute for Institute of Health Economics and Clinical Epidemiology of the University of Cologne.

5.2 Unfamiliar area of study: The lack of scientific knowledge

Even within the medical community, knowledge and expertise in the area of rare diseases is lacking in many instances.

For example, immunology has only just been made a compulsory subject in the study of medicine. And yet, owing to the different structures of the medical degree in Germany, there is no uniformity in the nationwide training of doctors.

Most doctors encounter a rare disease only once during their working lives, if at all. Additionally, there is too little research conducted in Germany into rare diseases:

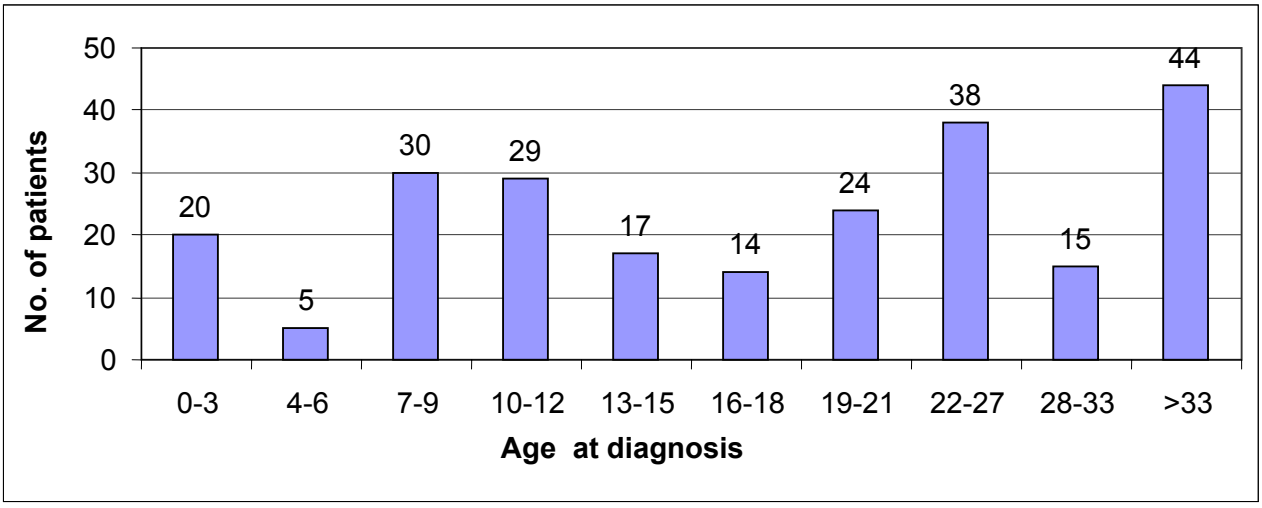
> *“France spends one hundred times the amount of money on genetic research and on the study of rare diseases. The establishment of special research centers and programs in Germany is necessary for science and very important for public awareness and politics.”*
> Prof. Dr. med. Reinhold E. Schmidt, Department of Clinical Immunology, Center for Internal Medicine of the Medizinische Hochschule Hannover, President of the German Society for Immunology (DGfI)

Therefore, the knowledge of students, practicing doctors and scientists in the areas of immunology and rare diseases must be improved as a matter of urgency.

5.3 Lost chances: Lack of diagnosis ▲

There is a significant lack of diagnosis of rare illnesses.

Many sufferers frequently obtain a correct diagnosis only after consulting multiple doctors over a period of years. Others are never correctly diagnosed. Either the correct diagnostic procedure is not followed, or doctors are simply unaware of one.



Statistical evaluation of membership of the age at diagnosis, DSAI e.V.

Where a diagnosis comes late or is incorrect, patients with rare diseases cannot be effectively treated. In addition, there is the risk that unnecessary treatments will be performed based on misdiagnoses. A broad-ranging study with nearly 6000 participants Europe-wide revealed that 40% of sufferers of 8 rare diseases were initially wrongly diagnosed and 16% of these were operated upon on the basis of the misdiagnosis.[9]

> *“The importance of the early recognition and treatment of primary immunodeficiencies in preventing further harm is proved by an Italian multicenter study from 2002, which reveals that in the case of one of the most severe antibody deficiency syndromes, X-linked agammaglobulinemia, a delayed diagnosis, e.g. not until the age of 10, led to irreversible lung damage in 40% of children.”*[10]

[9] Eurordis-Studie “EurordisCare 2: Survey on the delay of diagnosis for 8 rare diseases in Europe”, 2004.
[10] Plebani, A./Soresina, A./Rondelli, R. et al.: Italian Pediatric Group for XLA-AIEOP. Clinical, immunological, and molecular analysis in a large cohort of patients with X-linked agammaglobulinemia: an Italian multicenter study. Clin. Immunol. 2002.

An early diagnosis is critical to successful treatment. Where there is timely and adequate treatment, patients with primary immunodeficiency, for example, have an almost equal life expectancy to that of the population average.

5.4 (Mis)treatment: Inadequate medical care ▲

There are further inadequacies in the areas of treatment and patient care.

Frequently, effective treatment is not given because the actual causes of the illness are unresolved. The rarer the illness, the more systematic research is made difficult.

As the illness or the resulting complications of an undiagnosed illness frequently affect multiple organs, interdisciplinary treatment approaches are necessary. Only a few specialist centers can provide these.

The example of primary immunodeficiency makes it clear that there must be a fundamental improvement in treatment facilities for sufferers of rare diseases.

> *“I suffer from a congenital immunodeficiency and have been treated at a university pediatric hospital in the immunodeficiency clinic in Munich since I was 2. This year I will turn 18, and become an adult. This means that the pediatric clinic will not be able to treat me any longer. Great – the nearest immunodeficiency clinic for adults is in Freiburg. This means I will have to travel hundreds of kilometers to receive good treatment.”*
>
> Mario Gründl, sufferer

The treatment situation is particularly worrying for adult patients with primary immunodeficiency. Currently, there are only a few immunodeficiency centers nationwide. The adult treatment center in Munich, for example, was closed.

> *“Currently in Germany there are only about four or five treatment centers for adult patients with immunodeficiency. Often treatment is not approved owing to the uncertainties of the insurance providers and the health fund situation. “*
>
> Prof. Dr. med. Reinhold E. Schmidt, Department of Clinical Immunology, Center for Internal Medicine of the Medizinische Hochschule Hannover.

Owing to the low number of adult immunodeficiency centers, adult sufferers are treated by pediatricians at great cost:

> *“In Berlin, pediatricians even treat 80-year-old sufferers and incur significant costs: per quarter and patient a clinic receives 50 Euro from the health funds, when costs can be up to 3000-4000 Euro. Thus there is great pressure upon the clinics to stay afloat or close.”*
>
> Prof. Dr. Volker Wahn, Campus Virchow-Klinikum Berlin, Clinic for Pediatrics, specializing in Pneumology and Immunology.

The direct results of the dire treatment situation include high travel costs and absence from work for sufferers and the enormous costs and burdens placed upon the specialist centers. Nationwide treatment for all sufferers must be ensured through the provision of immunodeficiency centers.

5.5 Between a rock and a hard place: Treatment costs ▲

In the area of treatment costs, the example of primary immunodeficiency also highlights the difficult situation faced by patients with rare diseases: sufferers of primary immunodeficiency are classified as chronically ill, yet they are not able to be automatically classified under the practice budget of their treating doctor as an exemption. Doctors must either deal with the large amount of bureaucracy necessary to have a PI sufferer classified as a practice exemption, or they have to accommodate their treatment within the existing practice budget. Yet the latter then results in many patients not receiving sufficient treatment for cost reasons. The resulting “stop-gap” treatment, however, is insufficient and cannot, for example, clinically stabilize primary immunodeficiency patients. This results in recurrent infections, despite treatment.

Such short-term, cost-saving measures are, in effect, highly counterproductive when considered in the light of the expense of the intensive treatment of later serious infections and absence from work which this results. This in turn constitutes a further burden for health insurance funds.

> *“The chronically ill need to be made an exemption and removed from the budgetary constraints of the doctor’s practice.”*[11].
>
> Gabriele Gründl, President, German Self-Help Organisation for Congenital Immunodeficiencies e.V.

5.6 Isolation: Social consequences ▲

An undiagnosed or insufficiently treated rare disease impacts upon all areas of life: school, career, leisure and emotional wellbeing. It can lead to isolation and exclusion from society; to lack of understanding in the workplace and even amongst doctors; to diminished career chances and financial discrimination from health insurers.

At least one of these consequences can be avoided. When patients spend years without a proper diagnosis, they lose their trust in the health care system and their treating doctors. So consequences such as an inability to work and early retirement could be avoided in many cases by adequate and appropriate treatment.

It is not only the person with the rare disease that faces lifelong suffering – their family is also affected. Many parents blame themselves, relatives experience the helplessness and

[11] Patientenbrief, Ihr gesundheitspolitischer Fakten- & News-Letter, GSK, May 2006.

uncertainty of overburdened practitioners. Frequently, parents separate after the birth of a child with a rare disease.

Experiences:

> *“Ever since I was very young I have always been sick. When I was 4 months old I developed pneumonia and as a child frequently had bronchitis which, however, was able to be cured…But then it really started when I was thirty, and things became serious.*
>
> *As an adult, a doctor advised me to have the window in my nasal sinuses enlarged. So I underwent the operation, but this didn’t help. I caught bronchitis again, and the pneumonia did not go away. My illness isolated me. I didn’t go to large events for the fear of infecting someone. No concerts, no family parties. The lowest point of my social isolation came when I had to give up my job as a teacher. Work at the school had become unbearable for me. I contracted anything that the students had. Each harmless cold that student had meant certain severe bronchitis for me.*
>
> *When I was 50 I was admitted to a hospital in Siegen with suspected lung cancer. I underwent many tests. Marrow was taken from my pelvic bone. I couldn’t stand not knowing what was wrong. I was afraid and I did not want to die. But it was worse seeing the fear in the eyes of my wife and children. They had suffered for years alongside me. Then a message came from the oncology department: “Mr Wulfleff, we know what is wrong with you…” “You are suffering from a rare disease: antibody-deficiency syndrome, a congenital immunodeficiency. Your levels of IGG and IGA are practically negligible. But it can be treated, and we can help you get back to leading a normal life.” It was hard to grasp: I had lived with an unknown illness for 50 years and finally a doctor had established what it was…”*
>
> Extract from Harald Wulfleff’s account of his illness

> *“Hi, I am Moritz and I am eleven and am in the fifth grade. Sport is my favourite hobby. I am good at rollerblading bike riding, skateboarding and I play football, proper football, in a team. Oh yeah, and I have a congenital immunodeficiency.*
>
> *When I was little, my parents kept having to take me to the doctor. I was only one or two…I was sick all the time, and I had bronchitis and other things. We were always seeing the pediatrician, because I had quite a cough. He gave me medication, antibiotics, I think. When we were home again, the cough got better, and I was well for a few days. But then the infection came back. And that was always how it happened. Moritz has bronchitis, Moritz goes to see the pediatrician, the doctor prescribes antibiotics, Moritz gets better for a little bit – and Moritz then was sick again after a few days. That was really dumb.*
>
> *…Then, when I was two-and-a-half, I got pneumonia. That was bad and it really hurt. I was sent to the pediatric hospital. The doctors did many tests on me, took some blood…Even though I was still very little, I still noticed that there was something wrong with me, that I wasn’t healthy. And even though everyone knew that I was sick, the doctors couldn’t find out what was wrong with me exactly. So I had more antibiotics to take, the pneumonia got better and I was able to go home again…*
>
> *A little while later I really had to go back into hospital – pneumonia again…I had to go to the place where the really sick people were – into intensive care. Finally the doctors tested my blood to find out about my immune system and they found out that it was in a bad state, because I didn’t have any antibodies. “Your son has a congenital immunodeficiency” was what the doctor said…The doctor then explained to me that my blood has lots of troops in it who defend against invaders, such as flu viruses. He said that in my body, some of these troops are missing and that’s why I always get sick. I got the gist of that. And then the doc said to my parents “There are successful treatments for this today. We can help your son…”*
>
> Extract from Moritz Sommer’s account of his illness.

> *“…I had just turned five when the problems began. I was playing and suddenly felt sick. From this day onwards, my life was marked by visits to the doctors. I can remember how I always had to take antibiotics for middle ear infections, sinus infections, suppuration and bronchitis. Taking pills became routine for me, and*

they became a kind of basic food. My mother had to spend many nights sitting at my bedside, because I couldn't sleep for all the pain…

When I began school in September 1973, my bronchitis reached its worst. Coughing, fever and a runny nose became my constant companions. During this time, I wasn't allowed to go to school, the teachers found my constant coughing a disturbance. I had many days off, and for years no-one knew what was wrong. The diagnosis – congenital immunodeficiency (hypogammaglobulinaemia) – was made in June 1981…

I have been treated by Prof. Metzner in the Immunology Department of the University Hospital in Leipzig since September 2005. Unfortunately, he is about to retire, and I am still not sure what will happen after this. Unhappily, I have had constant problems with other doctors, who did not want to prescribe me the immunoglobulin I required. They would tell me things like "I can't do anything for your condition – you will have to find a specialist", or "Stay put in the specialist clinic, they have a bigger budget". Or even "Well I can't prescribe this medication – my budget won't cope with that." In answer to questions such as "Should I start looking for a place to be buried?" came answers such as "Yes, you might as well do that." My earlier ear nose and throat doctor used to say to me "Another holiday?" each time he handed over a prescription. Of course I changed doctors, but in emergencies you don't have a choice but to take the doctor who is on duty at the time. Recently I have been taking my medical results from Leipzig with me, so that doctors can see straight away what the problem is, should there be an emergency. But most of the time the doctors do not have a clue how they should treat me and I mostly have to determine what the treatment is."

Extract from Annette Olboeter's account of her illness.

6. Rare diseases – A challenge for the health care system ▲

6.1 Today's costs – Tomorrow's savings ▲

The neglect of rare diseases incurs substantial financial and social costs for the German health care system, taxpayers and sufferers due to:

a. late and/or incorrect diagnoses
b. incorrect treatment
c. work disability and early retirement

Through early diagnosis, expensive and unnecessary, incorrect treatments could be avoided. Early diagnosis can be made with minimal financial costs. The cost of a primary immunodeficiency basic diagnostic procedure is only 3.60 Euro.

Where a diagnosis is lacking, it is nearly impossible for sufferers to perform regular work, owing to the constant infections and their irreversible complications. The consequences of this are early retirement, impoverishment and social regression. In addition, when sufferers are no longer able to work, they are also no longer able to contribute as taxpayers to the welfare system.

A reevaluation of the diagnosis and treatment of rare diseases is thus of vital importance for sufferers and for the health care system.

A coherent approach to rare diseases can be decisive in the long-term improvement of the cost-efficiency of the health care system. Hence the speedy implementation of **basic measures** by

the Federal Government, the Federal Parliament, the Federal Joint Committee, representatives of the health insurance funds, doctors' bodies and hospitals is urgently needed.

6.2 Action now: Essential measures ▲

6.2.1 Education of policy makers and the public about rare diseases

A clear increase in the awareness and information levels among policy makers and the public about the difficult medical and social situation of people with rare diseases, and particularly primary immunodeficiency.

"The stigma attached to rare diseases must be removed."

Prof. Karl W. Lauterbach MdB, Institute for Institute of Health Economics and Clinical Epidemiology of the University of Cologne.

6.2.2 More efficient integration of rare diseases with the health system reforms

Give greater prominence to rare diseases, in particular primary immunodeficiencies and improve their integration within the health care system. This is an important step to be taken in conjunction with health system reforms.

6.2.3 Improved cooperation between policy makers, science, industry, researchers and practicing doctors.

Greater cooperation between policy makers, science, industry, researchers and practicing doctors to guarantee the best possible treatment for sufferers.

"And precisely with the example of rare diseases, it becomes clear that politics, science, research, medical practitioners, industry and those affected must all cooperate intensively to ensure the best level of medical care."

Helga Kühn-Mengel MdB, Federal Government Commissioner for Patients' Affairs

6.2.4 Education and better information for the medical profession

Make rare diseases and immunology a fixed part of the study of medicine and training of doctors. Expand training and further education programs for practicing doctors in the areas of rare diseases, so that patients can be diagnosed early and given adequate treatment.

"Specialist medicine requires a broad impact, intensive networking and an expansion of interdisciplinary treatment."

Emilia Müller, Bavarian Minister of State for Federal and European Affairs

6.2.5 Early diagnosis for all sufferers

Make early diagnosis possible for all sufferers, so they have certainty and their quality of life can be improved.

“An unknown number of patients with primary immunodeficiency in Germany are often diagnosed too late and not adequately treated. This causes serious complications, which could be avoided with timely diagnosis and treatment.”

Prof. Dr. Volker Wahn, Campus Virchow-Klinikum Berlin, Clinic for Pediatrics specializing in Pneumology and Immunology

6.2.6 Guarantee proper treatment

All health funds should provide for an appropriate course of treatment in line with current medical standards to every patient with a rare disease. This should be made a reality, for the good of the patient and for the health system.

“If the diagnosis rate of primary immunodeficiencies does not considerably improve, then the long-term treatment costs will increase owing to misdiagnosis. An early diagnosis is clearly more cost effective.”

Prof. Dr. med. Reinhold E. Schmidt, Department of Clinical Immunology, Center for Internal Medicine of the Medizinische Hochschule Hannover, President of the German Society for Immunology (DGfI)

6.2.7 Ensuring nationwide medical care with specialist clinics and centers for treatment and research.

An adequate network of contact points must be guaranteed for adults and children alike, where specialists are available for diagnoses and treatment.

“Every patient rightfully expects that all diagnostic and treatment possibilities will be exhausted in order to offer them sound medical care, irrespective of their income or level of education.”

Helga Kühn-Mengel MdB, Federal Government Commissioner for Patients’ Affairs

6.2.8 Inclusion of treatment costs within the rebate policies of health funds

A comprehensive legal framework to ensure compulsory inclusion of treatment costs for rare diseases in the rebate schemes of the health insurance funds.

“The chronically ill need to be made an exception and removed from the budgetary constraints of the doctor.”[12]

Gabriele Gründl, President, German Self-Help Organisation for Congenital Immunodeficiencies e.V.

[12] Patientenbrief, Ihr gesundheitspolitischer Fakten- & News-Letter, GSK, May 2006.

7. The “Rare diseases – Not at all that rare” campaign and the Berlin Signal

7.1 Our objectives ▲

Our campaign wants to ensure that the topic of **rare diseases** occupies a **more prominent place** on the **political and public agenda** and to achieve a **reevaluation of diagnostic and treatment techniques**. Our **objectives** are:

- To guarantee all sufferers the provision of early diagnosis and adequate treatment in accordance with current medical standards.
- A fundamental improvement in the quality of life for sufferers.
- To increase the long-term cost efficiency of the health system by avoiding both costly or incorrect treatments and the high social costs incurred through work disability and early retirement.

7.2 Our call for action: Catalogue of concrete measures ▲

To achieve our goals we are calling for the implementation of three concrete measures:

7.2.1 Guarantee of finance for contracts under § 116 b, sub. para. 2, 3 Sozialgesetzbuch V (German Social Security Code V)

Under this law, special contracts can be made between hospitals and health funds to provide additional outpatient treatment in the hospital. This applies in cases involving “highly specialized services, rare illnesses and illnesses with a particular etiopathology”. It includes the “diagnosis and treatment of patients with severe immunological illness”, such as antibody-deficiency syndrome.

Because this law does not currently mandate such a practice, health insurance funds have yet to agree to finance such a contract, as this would mean they would then have to provide specific funds in addition to the existing budget for in- and outpatient treatment. This leads to the under-funding of specialist centers and to inadequate treatment, particularly of adult patients with rare diseases.

Therefore, health insurance schemes must be obligated to conclude special contracts with hospitals, upon request, for the outpatient treatment of rare diseases, in order to redress the inadequacies in treatment. The financing of such contracts needs to be guaranteed.

7.2.2 Classification of replacement therapies as a practice budget exemption in the case of antibody-deficiency syndrome.

Adequate treatment of patients with primary immunodeficiency in established practices often fails on account of the fear on the part of doctors that they will

have to cover the costs of expensive medication therapies out of their own fees, with their budgetary guidelines having been exceeded.

Therefore, treatments involving medications for primary immunodeficiency should be included in the catalogue of practice budget exemptions under budgetary spending agreements between doctors and insurers. In this way, the additional financial requirements of particular therapies would be exempted from the normal budgetary guidelines and the expenses incurred for the medications would not place an additional burden on a practice.

7.2.3 Inclusion of screening for antibody deficiencies in the statutory preventative medical checkup of infants and young children / “Yellow Book”

The inclusion of screening for antibody deficiencies in the preventative medical examination of small children with severe infections as well as the inclusion of a blood count would ensure early detection and diagnosis.

We call for the following changes in the standard preventative medical checkup recommended by statute, in order to improve screening for congenital immunodeficiencies:

In **U2** and **U3** a new box under “REQUESTED INFORMATION”:
Cosanguinity, infant/child deaths in family.

In **U3** and **U4** (4-6 weeks.; 3-4 months) under “CLINICAL FINDINGS”:
Chronic inflammatory changes in skin or signs of a graft-versus-host reaction (affecting inside of hands, sole of feet)

U4, **U5**, **U6** (4-6 weeks.; 3-4 months, 10th-12th month), **U7** (21st-24th month) and **U8** (4 years) under “REQUESTED INFORMATION”:
Instead of “cumulative infections” (current version), replace with “pathological susceptibility to infection”.

7.3 Berlin Signal ▲

Our campaign goals, demands and catalogue of concrete measures to improve the treatment situation of sufferers in Germany are summarized in our **Berlin Signal**.

The **Berlin Signal** forms the central core document of our current campaign phase and should serve as a basis for discussions within the policy debate in the area of public health care.

In conjunction with the **Berlin Signal** we are conducting a signature campaign to gather support for our demands. The **Berlin Signal** can be downloaded at http://www.berliner-signal.de and supporters can add their signatures there in electronic form.

8. Conclusion

Patients with rare diseases currently exist on the margins of the health care system, often without a correct diagnosis and appropriate treatment.

The urgent problems posed by rare diseases, in particular primary immunodeficiency, must be given greater attention within the political and public spheres. Rare diseases pose an enormous challenge for the health care system.

Early diagnosis of rare diseases brings improvements to the health and quality of life of sufferers and benefits the health care system, in that unnecessary and inappropriate treatments can be avoided.

A late diagnosis or a lack of diagnosis, on the contrary, leads to high social costs: the potential inability to work and early retirement from the workforce.

Political decision makers and all relevant health care institutions have the common duty to ensure that the treatment situation of sufferers is made the best it can be and to use every chance to improve cost efficiency.

A pooling of medical, political and scientific resources is urgently needed to provide systematic research, optimal exchange of information and ideas and the lasting groundwork for early diagnosis and appropriate treatment to achieve improved patient care.

Providing an effective health care system and ensuring the wellbeing of patients within it are the common responsibilities and duties of policymakers, business and industry and civil society.

We urgently call upon the Federal Government, the Federal Parliament and all relevant health care policy institutions to study our catalogue of measures, to discuss them and to integrate them in an appropriate form into German health care policy.

BERLIN, 4 September 2006

9. Further information ▲

Further information can be found under the following links:

National and international patients' organisations and networks:

http://www.dsai.de
(German Self-Help Organisation for Congenital Immunodeficiencies - DSAI e.V.)

http://www.achse-online.de
(Alliance for Chronic Rare Diseases - Achse e.V.)

http://www.eurordis.org
(European Organisation for Rare Diseases)

http://www.ipopi.org
(International Patient Organisation for Patients with Primary Immunodeficiencies (IPOPI)

http://www.efis.org
(European Federation of Immunological Societies)

http://www.esid.org
(European Society for Immunodeficiencies – ESID)

http://www.orpha.net und http://www.orphanet.de
(European Database of Rare Diseases)

http://www.immunologie.de
(German Society for Immunology)

http://www.jmfworld.org
(Jeffrey Modell Foundation)

In the European Union

http://ec.europa.eu/dgs/health_consumer/index_de.htm
(EU Commission, D-G Health and Consumer Protection)

http://ec.europa.eu/health-eu/index_en.htm
(EU Health Portal)

http://ec.europa.eu/health-eu/health_problems/rare_diseases/index_en.htm
(Features a section on rare diseases)