

Programme Review 2012

RARE DISEASES

Research Going Beyond

ALL RESEARCH PROJECTS AT A GLANCE Page 6

A DOCTOR? NO. I WANT TO BE A COOK. Page 2

WE CANNOT ALWAYS DELIVER
THE GOLD STANDARD. Page 4

A NATIONAL STRATEGY IS NEEDED. Page 8

THE PHARMACEUTICAL INDUSTRY
IS MORE INTERESTED IN RARE DISEASES TODAY. Page 10

NETWORKING. BUT HOW? Page 12

GRSTIFTUNG.CH

— GEBERT RÜF STIFTUNG —
WISSENSCHAFT.BEWEGEN

RE(ACT)[®]

INTERNATIONAL CONGRESS ON RESEARCH OF RARE AND ORPHAN DISEASES
29 FEBRUARY – 02 MARCH 2012
GEHRY BUILDING, BASEL, SWITZERLAND, REACT-CONGRESS.ORG

Orphan diseases in Switzerland: Things are gradually moving

Rare diseases are actually a common disease, because although for each rare disease there are relatively few patients, roughly five percent of the population are affected by rare diseases, since there are about 7000 different diseases. The definition is: A disease that occurs less than once per 2000 citizens is considered to be rare. There are rare diseases of which only a handful of cases worldwide are documented.

For many years rare diseases were considered to be the orphans of medicine. Most doctors and most pharmaceutical companies had only little interest in them. But in recent years, there has been some progress in Switzerland at many levels: research groups have been constantly achieving new advances in diagnosing and treating rare diseases, in the pharmaceutical and biotech industry the subject has moved higher up the list of priorities, the interest of health care politicians has increased, demand for a national strategy is clearly audible, and on the part of patients and patient groups there is noticeable optimism.

Gebert RUF Stiftung is supporting research projects in the area of rare diseases with approx. ten million francs to be allocated over five years. The initiative aims at developing and implementing innovative technologies and approaches in the diagnosis and treatment of rare diseases.

Rare Diseases News

WE WILL SOON MEET: RE(ACT) CONGRESS 2012 ON RESEARCH OF RARE AND ORPHAN DISEASES IN BASEL

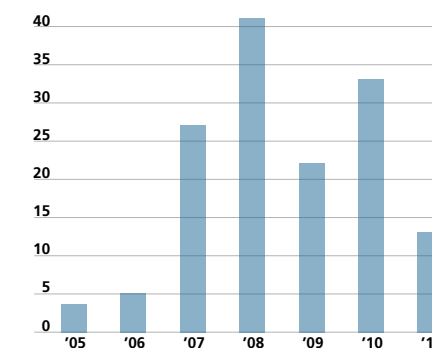
From 29 February to 2 March 2012 the first International Congress on Research of Rare and Orphan Diseases will take place at the Gehry Building located on the Novartis Campus in Basel. RE(ACT) will bring together world leaders and young scientists from universities and industry to present cutting edge research, to discuss results and to exchange ideas. The congress also aims at promoting research on rare and orphan diseases among the general public, industry and policy makers, and at helping to foster an understanding of other more common diseases and to encourage clear insight positions. The main research topics to be discussed at the congress will focus on gene and cell therapy, stem cells, diagnostics, therapeutic applications and genomic disorders.



Registration is open until 31 January 2012. On-site registrations will be accepted subject to available places. The congress was initiated by the BLACKSWAN Foundation and Gebert RUF Stiftung's "Rare Diseases, New Approaches"-Programme and is supported by Eurordis (Rare Diseases Europe), ProRaris (Swiss Rare Disease Alliance) and E-Rare (ERA-Net). react-congress.org

GOOD NEWS: THE NUMBER OF ORPHAN DRUGS IN SWITZERLAND IS RISING

The number of drugs with orphan drug status in Switzerland has increased considerably since 1 January 2010. All in all, there are currently 143 indications from 105 drugs (as per the end of July 2011). Since the start of 2010, Swissmedic has given the go-ahead to 45 new indications.



Indications with orphan drug status in Switzerland (as per end of July 2011)

The orphan drug status conferred by Swissmedic means that the manufacturers of these products are able to benefit from a simplified registration procedure. This new status was introduced in 2006. Allowance is made, inter alia, for the fact that, with rare diseases, the number of test persons is limited, making it more difficult to conduct studies. This is why drug manufacturers can also submit dossiers which have not been fully completed to Swissmedic and do not need to pay any fees for the appraisal. swissmedic.ch

DIAGNOSIS FIRST: UNDIAGNOSED DISEASES PROGRAM

To address the fact that there are still many yet undiagnosed rare diseases, in May 2008, the US National Institutes of Health (NIH) started the "Undiagnosed Diseases Program". To evaluate each patient enrolled in the new programme, NIH will enlist the expertise of more than 25 of its senior attending physicians, whose specialties include endocrinology, immunology, oncology, dermatology, dentistry, cardiology and genetics. The programme pursues two goals: To provide answers to patients with mysterious conditions that have long eluded diagnosis and to advance medical knowledge about rare and common diseases. More than 200 medical cases have been enrolled from among more than 1200 sets of patient records submitted by patients seeking answers to mysterious disorders.

In July 2011, because of the overwhelming number of applications received, the programme had to temporarily suspend accepting new applications for admission or new medical records. This year, the programme scored an initial success in discovering a novel vascular disease which the researchers refer to as ACDC, or arterial calcification due to CD73 deficiency. The adult-onset condition is associated with progressive and painful arterial calcification affecting the lower extremities, yet spares patients' coronary arteries. The rare arterial condition caused by calcium build-up in arteries below the waist and in the joints of patients' hands and feet has been observed in nine individuals from three unrelated families, who are the only people known to have the disorder.

rarediseases.info.nih.gov

Making gene therapy safer and more efficient

The research project being conducted under Gebert RUF Stiftung's Rare Diseases Programme by Janine Reichenbach and her group at Children's Hospital Zurich (Kinderspital Zürich) is attempting to make the gene therapy that was employed for Max more efficient and to modify it in such a way that it will have a longer-term effect. One problem with gene therapy is the gene shuttles that are used to deliver a copy of the healthy gene to the patient's sick cells. These are still not specific enough today and don't always reach the intended cells. And there is another problem too: if the gene shuttles target the wrong point in the genome, they can trigger cancer. "Our research project is aimed at finding better gene shuttles so as to make gene therapy safer and more efficient", explains Reichenbach. The project is also looking into a recycling mechanism for the immune cells, so-called macroautophagy.

A doctor? No. I want to be a cook.

Eleven-year-old Max still has a certain amount of difficulty walking, and his gait is stiff. This, however, is the only physical trace of those dark weeks seven years ago when he was lying in a bed in the Children's Hospital in Zurich, and the doctors were not sure whether he would ever walk again.

It started at the age of three months. "Max was always ill", explains his mother. First of all came the skin rashes, then serious bouts of pneumonia, and he regularly had a high temperature. His paediatrician conducted test after test, but came up with nothing. Children's immune systems could sometimes get mixed up, the doctor explained, the boy was basically healthy, he just had an infection.

When Max was two years old and there had still been no improvement, Max was referred to a specialist at a university hospital on account of inflamed lymph nodes. The specialist conducted a more detailed examination of the small boy and speculated that he was possibly suffering from a rare disease. Then, just a short time later, it was indeed established that Max had septic granulomatosis, a potentially fatal disease of the immune system. "We were lucky", says the mother, "it ,only' took two years for us to get the right diagnosis; it can take a great deal longer in other cases."

In granulomatosis patients, the immune system is weakened to such an extent that it can

scarcely stand up to pathogens. Any infection caused by a bacterium or fungus can constitute a fatal hazard for those affected. As of then, therefore, zoo and circus visits were taboo for Max, in the same way as swimming or excursions to the woods. The danger of him picking up a germ was just too great.

ONLY GENE THERAPY WOULD SAVE HIM

The disease can only be healed through a blood stem cell transplant, "but there was no suitable donor anywhere in the world, not even in the family", recounts the mother. Max was therefore first treated with different drugs to support his weakened immune defences, including antibiotics. Despite this, his health rapidly deteriorated and, at the age of four, Max became very ill: a fungus had attacked his lung and was advancing from there towards his spinal marrow. Within just two weeks, Max was paralysed. Only gene therapy would be able to save him. The therapy was a partial success, and the number of immune cells temporarily recovered to such an extent that they were able to overcome

the fungus. The doctors, however, were convinced that traces of the fungal attack would be left and that Max would probably never be able to walk again. His condition nonetheless improved, and after many months of therapy, he learned to put one foot in front of the other again.

Then, in 2009, there was new hope for Max: his little brother was born. This new life also served to give a new life to Max, since his little brother was a suitable donor for a blood stem cell transplant. In a further major procedure, Max's diseased immune system was destroyed, and then a healthy immune system built up again with the aid of blood stem cells from his little brother. For this, Max had to hold out for weeks under a plastic tent, isolated from the outside world and all the potentially fatal germs it held.

Today, his immune system is perfectly capable of warding off pathogens again, and he goes to school with his friends in the same age group. "My favourite subjects are maths and sport", the blond-haired boy proudly



declares. And he did not miss many lessons through his stay in hospital either, since he was able to take part in classroom teaching at home, via a webcam. And what does the future hold in store? "A doctor? No. I want to be a cook."

We cannot always deliver the gold standard.



Thorsten Hornemann (second from left) and his team at the Center for Clinical Research in Schlieren: Alaa Othman, Heiko Bode, Daniela Ernst, Glynis Klinke, Yu Wei and Iryna Sutter.

Thorsten Hornemann of the University of Zurich, one of Gebert Rűf Stiftung's grant recipients, on the difficulties of conducting a clinical study with patients who are suffering from a rare disease.

"A few years ago, a patient told me how he had realised for the first time that there was something not quite right with him", says Thorsten Hornemann, a metabolism specialist at the University of Zurich. The patient explained that he had been running over a pebble beach with friends. All the others were running because the pebbles hurt, like on a bed of nails, and he was the only one who was able to keep running without experiencing any pain. When he then looked at his feet, they were cut and bleeding. But he did not feel any pain. This patient is suffering from a particularly rare metabolic disease called HSAN1 (hereditary sensory and autonomic neuropathy, type 1). There are only 200 to 300 patients with this disease worldwide.

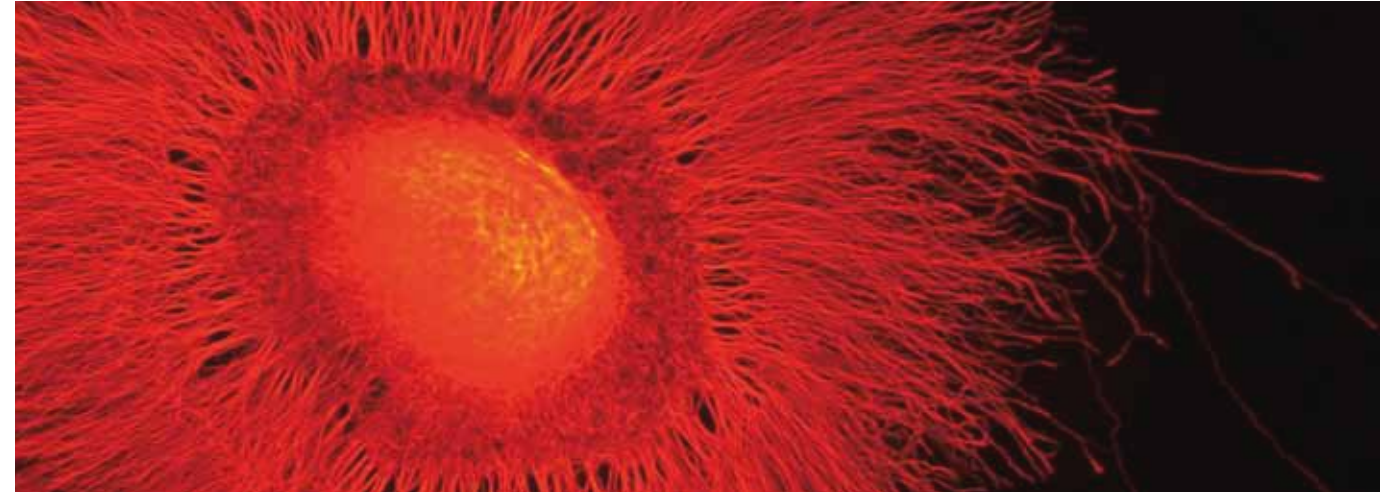
Due to a modified enzyme, a substance is produced in the cells of this patient's body that attacks the nerve cells. As a result, the axons – the connections between the nerve cells – become shorter, and the transmission of pain signals from the feet to the brain is interrupted. This generally starts at the age of around 16 to 20 years, first in the feet, before potentially spreading to the arms and finally to the entire body. The illness is not life-threatening, but repeated obstinate inflammation occurs and, in many cases, amputations may be necessary, such as of toes.

NO TREATMENT – UNTIL RECENTLY

"There was no treatment – until recently", explains Hornemann. His research group was able to show on mice to begin with that administering the amino acid serine can alleviate the illness. In order to test whether these results could also be transposed to humans, a small pilot study was conducted with a total of 14 patients with HSAN1. These patients took serine on a daily basis for ten weeks – with encouraging results: even after this short time, the patients' symptoms had improved.

"It is now a matter of confirming the results of the pilot study in a bigger clinical study lasting around two years", says Hornemann. He and his team then find themselves confronted with the typical hurdles facing researchers wishing to conduct a clinical study with patients suffering from a rare disease: there are only a few patients and these are generally spread all over the globe, making it difficult to obtain uniform measurement results. In addition, placebo-controlled, double-blind studies are regarded as the gold standard in medicine – but it is frequently impossible to conduct such studies at all in the case of rare diseases.

To take things in turn: in order to establish whether the serine therapy works, it is necessary to precisely record the state of health of



Picture of a nerve cell, starting to produce axons

each individual at the start of the study. Only in this way is it possible to ascertain whether there has been any improvement of medical relevance. These examinations cannot be performed by the patient's general practitioner but only by a specialist. This guarantees the uniformity of the data but does, however, mean that a patient from Scotland has to travel to London to be examined there. "The patients are fundamentally interested in taking part in studies, but the more travel expenses a patient has to pay, the lower their readiness to participate in a study will be", explains Hornemann.

A further challenge is eliminating the placebo effect: The doctor does not know whether he/she is giving the patient a placebo or the active ingredient, and the patient does not know if he/she is receiving a placebo or not. "In our case this is virtually impossible, because a patient can find out relatively easily whether they are being given a placebo or not", explains the metabolism specialist. In the case of HSAN1 patients taking serine, for example, their finger nails undergo a change,

becoming thicker. This clearly indicates to the patient that they are receiving serine and not a placebo.

Another obstacle is encountered with the "cross-over". In many cases, the patient groups are switched halfway through the study. The placebo group is then given the active ingredient, and the group that received the active ingredient is given the placebo. This enhances the information value of the study. The small number of patients with a rare disease means that a "cross-over" of this type is frequently impossible in clinical studies on rare diseases.

All these obstacles have an impact on the registration and, ultimately, on the payment of the cost of the drugs, since the registration authorities as well as the health insurance schemes wish to have sound data on which to base their decision as to whether to accept an active ingredient and assume the cost of a specific treatment. Registration authorities and health insurance schemes want the gold standard. "But we are frequently unable to deliver this gold standard in the case of rare

diseases, which is one of the reasons why there are only a few specific drugs for rare diseases, and why the costs of treatment are not paid", explains Hornemann.

Supported Projects

Via project data base descriptions and results are accessible: grstiftung.ch

Call 2010: 5 out of 48

2011–2013



Friedreich's Ataxia

Ass. Prof. Dr. Marc Bühler, Friedrich Miescher Institute, Focal Area Epigenetics
Funding: CHF 498 000

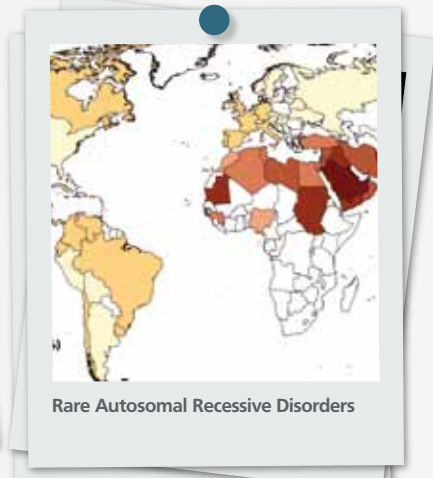
The precise mechanism of this disease of the nervous system remains enigmatic. The goal is to apply innovative technologies to gain insights into the pathogenic mechanisms and to search for novel therapeutics.



Chronic Granulomatous Disease (CGD)

PD Dr. Janine Reichenbach, University Children's Hospital, Department of Paediatrics
Funding: CHF 390 000

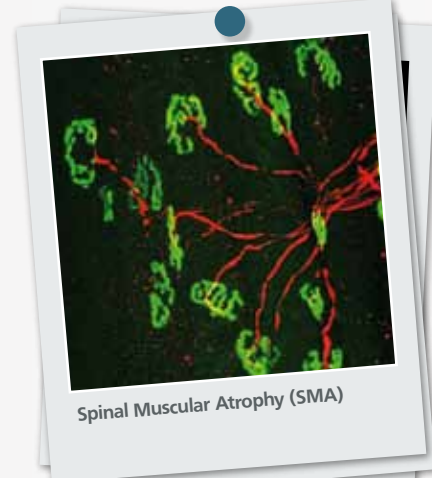
CGD can be cured by using gene therapy. This research project is attempting to improve gene therapy, making it more efficient and modifying it in such a way that it will have a longer-term effect.



Rare Autosomal Recessive Disorders

Prof. Dr. Stylianos Antonarakis, University of Geneva, Division of Medical Genetics
Funding: CHF 500 000

This study endeavours to identify mutations in the genomes of affected families and to introduce new diagnostic tests in the families with these rare disorders.



Spinal Muscular Atrophy (SMA)

Prof. Dr. Christoph Handschin, University of Basel, Department Biozentrum
Funding: CHF 400 000

SMA is a neuromuscular disease. This study has several aims, one of which is to find out whether the use of a certain protein (PGC-1alpha) has a therapeutic effect on the disease.



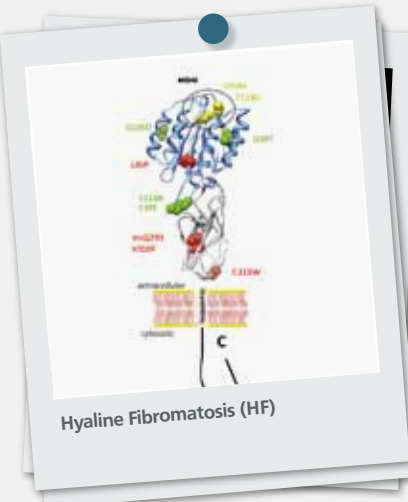
Lafora Disease (LD)

Dr. Oliver Kötting, Swiss Federal Institute of Technology Zurich, Institute of Agricultural Sciences
Funding: CHF 250 000

LD is a fatal genetic disorder for which there is no cure or treatment at the moment. This project wants to find the cause for LD.

Call 2009: 5 out of 58

2010–2012



Hyaline Fibromatosis (HF)

Prof. Dr. Gisou van der Goot, Ecole Polytechnique Fédérale de Lausanne, Global Health Institute
Funding: CHF 450 000

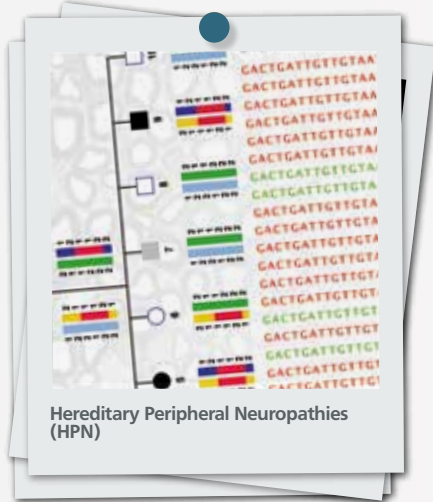
HF is a rare cancer. The purpose of this project is to better understand the composition of the tumors, identify cells that produce the hyaline material and try to find substances that could prevent tumor formation.



Polycythemia Vera (PV)

Prof. Dr. Radek Skoda, University Hospital Basel, Department of Biomedicine
Funding: CHF 300 000

PV is a rare leukemia. With the help of two families where this cancer occurs, the project wants to identify the genes that are responsible for the disease.



Hereditary Peripheral Neuropathies (HPN)

Dr. Carlo Rivolta, University of Lausanne, Department of Medical Genetics
Funding: CHF 440 000

HPN are rare disorders of the nerves. The study will hopefully lead to the identification of the gene mutations responsible for HPN.



Hereditary Sensory and Autonomic Neuropathy Type 1 (HSAN1)

Dr. Thorsten Hornemann, University of Zurich, Institute of Clinical Chemistry
Funding: CHF 340 000

The intake of the amino acid serine seems to help patients suffering from HSAN1. This project wants to confirm this finding in a clinical study.



Rare Disorders of Sexual Development

Dr. Serge Nef, University of Geneva, Department of Genetic Medicine and Development
Funding: CHF 450 000

The goal of this project is to develop new genetic tests for individuals or families with disorders of sexual development.

Call 2011: 6 out of 32

2012–2014

Rare Colonic Diseases

Prof. Dr. Jean-Christoph Leroux
Swiss Federal Institute of Technology Zurich, Institute of Pharmaceutical Sciences
Funding: CHF 300 000
The research team wants to develop novel biomolecules (peptides) that will be used to treat rare colonic diseases.

Dysferlinopathies

Prof. Dr. Michael Sinnreich
University Hospital Basel, Departments of Neurology and Biomedicine
Funding: CHF 480 000
This project includes a proof-of-principle clinical trial with the already approved drug Bortezomib in patients suffering from Dysferlinopathies (muscle wasting).

Inflammatory Bowel Disease

Prof. Dr. Anne Müller
University of Zurich, Institute of Molecular Cancer Research
Funding: CHF 190 000
It seems that infection with the gastric bacterium *Helicobacter pylori* helps to protect from inflammatory bowel diseases. This project wants to scrutinise this hypothesis.

Chronic Mucocutaneous Candidiasis (CMC)

Prof. Dr. Salomé Leibundgut
Swiss Federal Institute of Technology Zurich, Institute of Microbiology
Funding: CHF 500 000
This project wants to improve the understanding of the molecular mechanisms leading to CMC with the aim of identifying novel therapeutic targets.

Lymphedema-Distichiasis

Ass. Prof. Dr. Tatiana Petrova
University of Lausanne, Division of Experimental Oncology
Funding: CHF 500 000
This rare form of a lymphedema is caused by a mutated protein called FOXC2. The research project wants to find new ways of treating the disease.

Prader Willi Syndrome (PWS)

Dr. Shivendra Kishore
University of Basel, Department Biozentrum
Funding: CHF 110 000
Understanding the molecular steps leading to PWS remains a crucial bottleneck in developing appropriate therapeutics. This project wants to apply new techniques to solve this riddle.

grstiftung.ch/en/portfolio.html



Design for a joint strategy in Switzerland

- a Swiss database for securing and disseminating knowledge
- support to ensure optimum cooperation between all the players involved
- creation of national competence centres
- improvement of European and international cooperation
- provision of equality of access to diagnosis and therapies
- improvement of fundamental research and clinical research

A national strategy is needed.

In December 2010, National Council member Ruth Humbel (CVP) submitted a postulate aimed at improving the health situation of persons with rare diseases.

“We have slipped behind other countries in this respect, to the detriment of the patients”, says Humbel. In March 2011, the National Council decided that there was indeed a need for action in this field, and took up the postulate.

Ms Humbel, what, in your opinion, are the biggest challenges facing patients with rare diseases?

Life is more difficult for these patients than for others, because it generally takes years for them to receive a correct diagnosis. The period of uncertainty drags on and, in the worst case, the patient is suspected of feigning illness, because the symptoms simply do not tally with any disease. In addition, large numbers of children are affected. As far as their families are concerned, this generally involves running

from one specialist to another over a period of many years, together with a nerve-wracking struggle to have the treatment costs paid for.

What is the situation at the political level, what are the biggest challenges there?

The problem is that we cannot really do justice to rare diseases with our canton-based health system. Our system already comes up against its limits with common ailments such as diabetes and cancer, and, with rare diseases, the weak points are exposed even more clearly, for the simple reason that national networking is particularly important in the case of rare diseases. “Rare” means that there are only a few patients – especially in Switzerland, which is already a small country to begin with. Networking at the level of patients, doctors and

service providers would concentrate the available knowledge, facilitate access to information and provide a basis for optimum treatment and care. In our current system, patients with rare diseases all too frequently fall between two stools.

Can you give us an example to illustrate these weak points?

Today, it can happen that one patient with a rare disease has a drug paid for by their health insurance scheme while another patient, who belongs to a different scheme, does not. Another example is the lack of registers in which the data on patients with rare diseases is recorded. This shortcoming applies across-the-board in the health system. A number of cantons, but not all of them, keep cancer registers, but these are not compatible with each other. This is just nonsense. We need a national strategy and national registers.

What would a strategy of this type mean for the hospitals?

There are approximately 7 000 rare diseases with just a few patients in each case – no hos-

pital is in a position to cover all of these. There ought to be just a few hospitals in Switzerland focusing on specific rare diseases and building up competences. International networking is also essential here.

Is this strategy also aimed at improving cost efficiency?

When it comes to cost efficiency, it is important to consider not only the price of a drug but also the cost of the entire chain of treatment, from the diagnosis to aftercare. The aim is not so much to reduce the costs as to deploy the resources more efficiently. This would create additional benefit for the patients.

The problem of rationing is especially important in the case of rare diseases, because drugs to combat rare diseases can be very expensive. What solutions would you propose here?

In my opinion, there is at least one positive side to the Swiss Federal Supreme Court’s judgement on Pompe’s disease, where the Court engaged in fundamental considerations concerning the maximum annual cost

of treatment and defined an upper limit of CHF 100 000 per year – namely, that this has set the discussion on rationing in motion. I regard it as wrong, however, for the Court to set a rationing limit. This question requires a political answer, and the criteria – which include the quality of life and life expectancy – must be defined by doctors and other medical experts.

parlament.ch (Postulat 10.4055)



“The pharmaceutical industry is committed to ensuring that Switzerland will draft a national strategy for rare diseases so that patients with rare diseases have good medical care and access to essential medicines. It is also necessary to implement regulatory incentives for research and development of diagnostics and orphan drugs, similar to the U.S. and the EU.”

Position of Interpharma, the association of Swiss pharmaceutical research companies, on rare diseases.

The pharmaceutical industry is more interested in rare diseases today.

Over the past few years, the subject of rare diseases has moved up the pharmaceutical industry's list of priorities. There are two main reasons for this shift: scientific and economic.

Since the human genome was decoded eleven years ago, knowledge of the relationships between genes and diseases has advanced enormously. The rare diseases have also benefited from this, since a large proportion are genetically conditioned. Rare diseases are often based on a relatively simple mechanism of action, which makes the development of a drug easier. The advances in diagnosis and the new options for examining patients for defects in their genome have provided completely new stimuli for the treatment of rare diseases. Today, research teams at universities and in industry have genuine prospects of being able to develop new active ingredients which will radically change the lives of patients with a rare disease. This is in contrast to many widespread diseases, where good progress

has been achieved in recent decades. These diseases, such as diabetes, are relatively easy to treat today but breakthrough are harder to achieve.

There are also economic reasons behind the industry's interest in rare diseases. One of these is the price of drugs, which is increasingly coming under pressure. However, newly approved drugs for so far untreatable diseases do not have to fight with generics. In addition, the development costs for a drug to treat a rare disease (orphan drug) can be lower, because the dossier that has to be submitted to the authorities for approval does not need to be so extensive. So the prospects to earn profits with orphan drugs have improved. And – according to the pharmaceutical industry – they would be even better if barriers for clinical trials and approval procedures were lowered.

THE MARKET WILL ALWAYS BE SMALL
One problem still remains, however: the market for orphan drugs will always be small on

account of the small number of patients, and the price per patient is correspondingly high – in Switzerland, up to CHF 500 000 per patient and year is paid in some cases. When, last year, the Swiss Federal Supreme Court mentioned an upper limit of CHF 100 000 on treatment costs for a rare disease, this caused a huge outcry. Specialists immediately criticised the Federal Supreme Court for not having paid nearly enough attention to the fact that the prices of drugs for widespread and rare diseases are not comparable with each other given the difference in patient numbers.

The industry is now speculating as to whether the Federal Supreme Court's decision will have any impact on the pharmaceutical companies' current commitment to research into rare diseases, because the industry rejects fixed cost thresholds. It is, after all, quite clear that a pharmaceutical company will at least wish to recover its research and development costs when developing a new drug. It is not certain whether this will still be possible with an upper limit of CHF 100 000.

A THRESHOLD VALUE IS HARD TO DETERMINE

There are two points that give rise to hope here. On the one hand, the Federal Office of Public Health immediately put the Court's decision into perspective, saying that it would still be necessary to establish in individual cases whether it was justified to pay the cost of orphan drugs or not and that a threshold value would be hard to determine. In addition, the industry is pursuing further goals when developing an orphan drug – it hopes to be able to extend its use to treat more widespread complaints. In this way, the development costs for the rare disease application could be more than recouped.

interpharma.ch/de/politik/Seltene-Krankheiten.asp (German and French)





Networking. But how?

In the field of rare diseases, loud calls for networking can be heard from all quarters: politics, science, health insurers, patient organisations and industry. Where does networking really make sense?

NETWORKING IN SCIENCE

When it comes to networking between research groups worldwide, scientists basically agree that the calls for cooperation do not make much sense here, since researchers generally already engage in networking themselves. This is particularly so in the field of rare diseases, given that there are usually only a small number of research groups that are specialised in a particular rare disease. "Networking between research groups can be left to the scientists themselves" is the conclusion of Thorsten Hornemann, a lecturer at the University of Zurich.

Pressure to engage in networking is also being exerted from the outside. Previously, researchers could perhaps still conduct a clinical study

with four patients and publish it in a specialist journal. This is virtually impossible today. Researchers now need to be networked in order to conduct good science.

What could be improved, however, is the networking of Swiss scientists in European or international research programmes, for example in E-Rare, a research programme on rare diseases under the Seventh EU Framework Programme.

NETWORKING OF PATIENT ORGANISATIONS

The situation is different when it is a matter of making headway with the problem of rare diseases at the level of society. Networking between patient organisations makes sense here. Switzerland has so far been lacking an organisation that can be perceived as a point of contact by the population and is able to highlight issues in the same way as the Krebsliga for cancer or the Schweizerische Diabetes-Gesellschaft for diabetes. An initial step in this direction was taken last year, when, on

26 June 2010, a total of 42 patient organisations set up the new umbrella association, ProRaris, which stands up for the interests of patients with a rare disease.

NETWORKING OF PATIENT DATA

Today, there is no national register for recording patients with the diagnosis of "rare disease" or "unknown – possibly a rare disease" Switzerland-wide. This means that neither the doctors nor the patients know of other patients with the same rare disease. This shortcoming is particularly serious in the case of rare diseases, since patients at times know more about their illness than the doctors and are in a position to help each other.

A national register of this type would also make it easier for doctors to find patients with comparable symptoms who are possibly suffering from the same rare disease. And it would make it easier to conduct clinical studies. Only through national and international networking will it be possible to find enough patients to set up clinical studies.

NETWORKING OF HOSPITALS

It goes without saying that if television doctor Gregory House actually existed, then there would be no need for networking between hospitals, because Dr. House is in a position to solve all his cases within a matter of days. But the reality is more complex, which is why networking between specialists is necessary for the correct diagnosis to be made. In the ideal case, that would mean the creation of a national competence centre for rare diseases: the proximity of the different specialists would then contribute towards reducing the number of patients with an unknown diagnosis. In Switzerland, several centers of competence are more realistic. They should focus on a defined field, for example rare diseases of the nervous system.

Research Going Beyond

Rare Diseases – New Approaches

With CHF 2 million p.a. the programme promotes applied research aimed at developing and implementing innovative approaches or technologies. The current initiative aims to improve the diagnosis and treatment of rare genetic diseases. The knowledge gained should lead to a better understanding of the genetic, molecular and biochemical processes underlying these diseases and pave the way towards new diagnostics and new forms of treatment.

Programme Goals

The call is aimed at researchers developing and implementing innovative approaches or technologies to address currently unresolved questions. A further aim of the programme is to improve the transfer of basic research findings into clinical practice. The focus must be on innovation, feasibility and effectiveness, while attaining high scientific and technological standards. The programme was established in 2009 as a five-year area of activity.

2012 Call

The 2012 call for projects will be launched in March 2012. The application deadline will be in July 2012. The call is open to researchers at Swiss universities, university hospitals, Federal Institutes of Technology, universities of applied sciences and to research institutions affiliated with Swiss universities.

Information

Dr. Pascale Vonmont, Deputy Director Gebert RUF Stiftung, pascale.vonmont@grstiftung.ch



Masthead

Gebert RUF Stiftung, Bäumleingasse 22/4, CH – 4051 Basel, tel. +41 (0)61 270 88 22, fax +41 (0)61 270 88 23, info@grstiftung.ch, grstiftung.ch

Concept and editorial: advocacy ag, Gebert RUF Stiftung; Graphics: aplus; Pictures: Christoph Läser, Photograph AG, Novartis