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Inhaltsverzeichnis	Seite

Das Thema der heutigen Zeitschrift sind „Seltene Erkrankungen“. Dass mindestens 6000 seltene Erkrankungen so gar nicht selten sind, ist auf dieser neuen personalisierten Briefmarke thematisiert.

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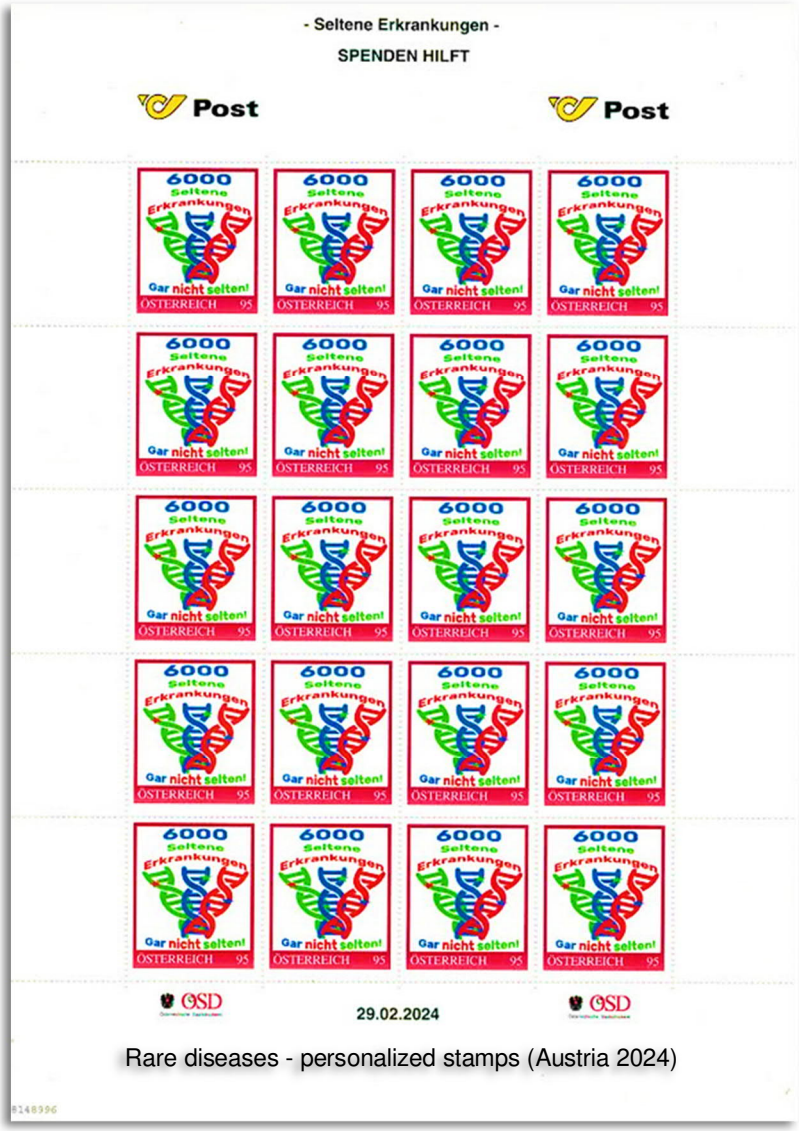
When you hear hoofbeats, think of zebras!

Daniela M. Vogt Weisenhorn



Rare diseases - personalized stamps (Austria 2024)

Around 70 years ago, Prof. Theodore Woodward gave his medical students the rule of thumb "When you hear hoofbeats, think of horses, not zebras". Since then, when making a diagnosis, many doctors have always thought first of the obvious and not of the unusual. However, what applies to common diseases (horses) can be misleading in the case of rare diseases (zebras) or they are not seen at all.

Rare diseases may have a low prevalence individually, but collectively they represent a significant challenge in the healthcare system. In the European Union, a disease is considered rare if it has a prevalence of less than one case per 2,000 people. It is estimated that there are 6,000 to 8,000 identified rare diseases worldwide.

The prevalence of individual rare diseases can vary from region to region and depends on various factors, including genetic predisposition, environmental factors and ethnicity. Some rare diseases have a higher prevalence than others, while many are extremely rare and only occur in a very small number of people.

It is important to note that although individual rare diseases are rare, the total number of people affected by a rare disease is considerable. In total, around 300 to 400 million people worldwide suffer from one of the identified rare diseases. This is symbolized on our new personalized ArGe stamp.

Regardless of the rarity of the diseases, some rare diseases are better known in our society than others. This is because they have received widespread public attention, be it through media reports, campaigns by patient organizations or well-known personalities affected by the disease. Some of these best-known rare diseases are:

ALS is probably known to many as the disease from which S. Hawking suffered (Isle of Man 2016)

- **Huntington's disease:** This neurodegenerative disease is known for its devastating effects on cognitive and motor function. It usually occurs in middle age and leads to progressive movement disorders (hence the former name St. Vitus' Dance), cognitive decline and psychiatric symptoms.

Modern dance can symbolize the "St. Vitus dance" (Lithuania 2023)

- **Amyotrophic lateral sclerosis (ALS):** ALS is a progressive neurological disease that affects the nerve cells in the brain and spinal cord that are responsible for controlling voluntary muscles. It leads to progressive muscle atrophy, paralysis and ultimately death due to respiratory failure.
- **Duchenne muscular dystrophy:** This genetic disease mainly affects boys and leads to progressive muscle wasting and weakness. Those affected often have difficulty walking, muscle cramps and heart problems.

Muscular dystrophy - special postmark Italy 2002

Cystic fibrosis - special postmark USA 1980

- **Cystic fibrosis (mucoviscidosis):** Cystic fibrosis mainly affects the lungs and digestive system and leads to chronic respiratory infections, digestive disorders and other complications. It is one of the best-known genetic diseases in children due to intensive (also philatelic) awareness campaigns.
- **Sickle cell anemia:** This genetic disorder affects the red blood cells and leads to an abnormal shape (sickle-like) of the red blood cells. This can lead to an increased risk of pain crises, anemia, infections and other health problems

Sickle cell anemia Awarness (Kenya 2016)

Diagnosis is also very important for tuberculosis - considered a rare disease in our country (Argentina 2014)

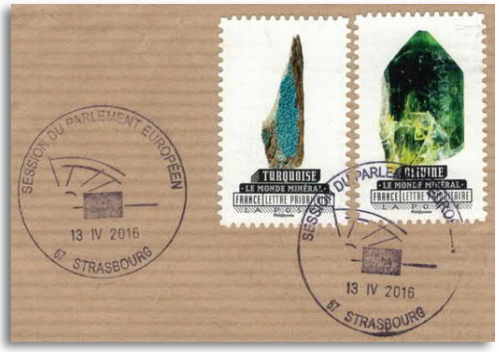
The challenges in the fight against rare diseases are complex. The first hurdle that patients have to overcome is diagnosis. Due to the limited experience and knowledge of doctors and the variety of symptoms, many patients go through an odyssey lasting years before an accurate diagnosis is made. For those affected, this not only means a considerable delay in treatment, but also emotional stress and uncertainty. In addition, most rare diseases are genetic in nature, which further complicates the situation. And even once a diagnosis has been made, it is not yet clear whether there is a treatment, as the development of therapies for rare diseases faces particular financial challenges, among other things. Due to the small number of people affected, the potential income from the marketing of drugs is limited, which often prevents pharmaceutical companies from investing in research and development. This leads to a lack of effective treatment options for many rare diseases.

But this is being counteracted by Regulation (EC) No. 141/2000, also known as the "Regulation on Orphan Medicinal Products" or "Orphan Drug Regulation". It is a piece of European Union legislation that aims to promote the development

Like Odysseus, patients have to go through many tests and a long journey before they are diagnosed. Like Odysseus, there are many tests before a diagnosis is made (Malta 2021)

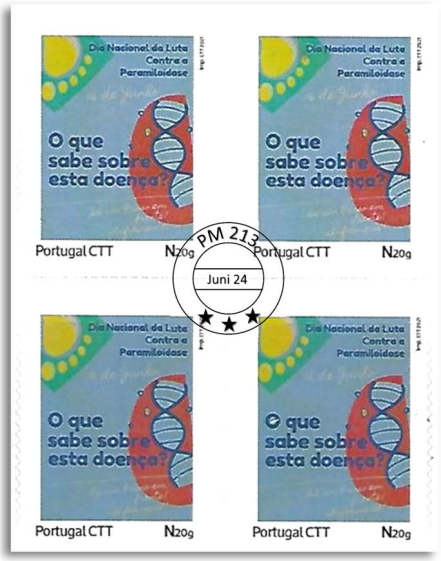
of and access to medicinal products for rare diseases, including the following points:

- Creation of market exclusivity: If a pharmaceutical company has developed a medicinal product for the treatment of a rare disease, no other company may bring an identical or similar medicinal product for the same indication onto the market for 10 years.



The "Regulation on orphan medicinal products" was adopted by the European Parliament (Strasbourg 2016 - stamp)

- - Creation of financial incentives: In addition to market exclusivity, companies that develop medicinal products for rare diseases can benefit from fee exemptions for regulatory procedures as well as financial subsidies and tax breaks.



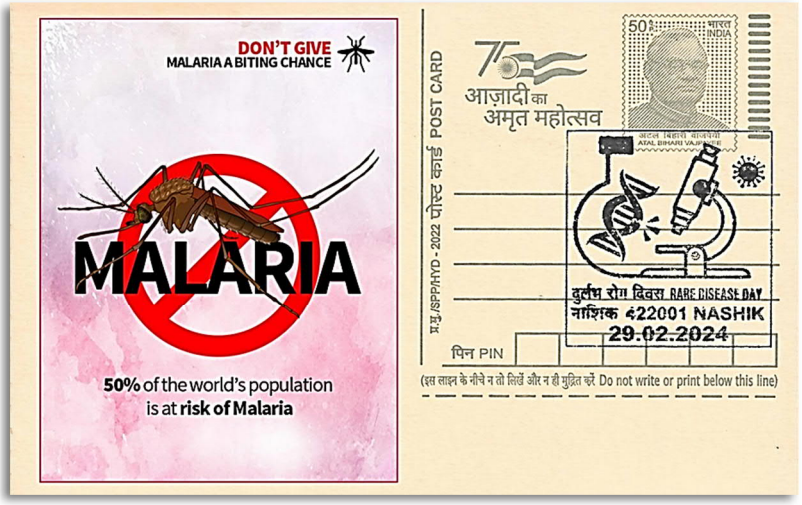
Raising awareness of the disease paramyloidosis through patient organization (personalized brand Portugal 2021)

International collaboration between researchers, physicians, patient groups and governments has also helped to raise awareness of rare diseases and mobilize resources for research and development. Initiatives such as Orphanet and Rare Diseases Europe (EURORDIS) play a crucial role in promoting research, education and support for rare disease patients.

However, the many patient organizations and self-help groups should not be underestimated. They provide important support for people with rare diseases and their families by offering information, resources, emotional support and the opportunity to exchange ideas with others affected. They also actively promote the interests of those affected and encourage research. To name just two of them: The German Society for Rare Diseases (SE Deutschland) and the Alliance of Chronic Rare Diseases (ACHSE e.V.). As a result of these initiatives, the Care Atlas for People with Rare Diseases (www.se-atlas.de) was created, which lists the addresses of clinics and organizations that those affected can turn to.

On a map of Germany you can find centers that deal with rare diseases (Germany 1994)

In summary, rare diseases represent a complex challenge that requires a coordinated effort at a global level. By raising awareness of these diseases, promoting research and development, improving diagnosis methods and access to treatment, the quality of life of people with rare diseases can be sustainably improved and they can be offered a more optimistic outlook for the future.



Raising awareness of rare diseases on "Rare Disease Day" with a special postmark (29.2.2024) on postal stationery from India. Interesting: postal stationery is dedicated to malaria, which is not a rare disease

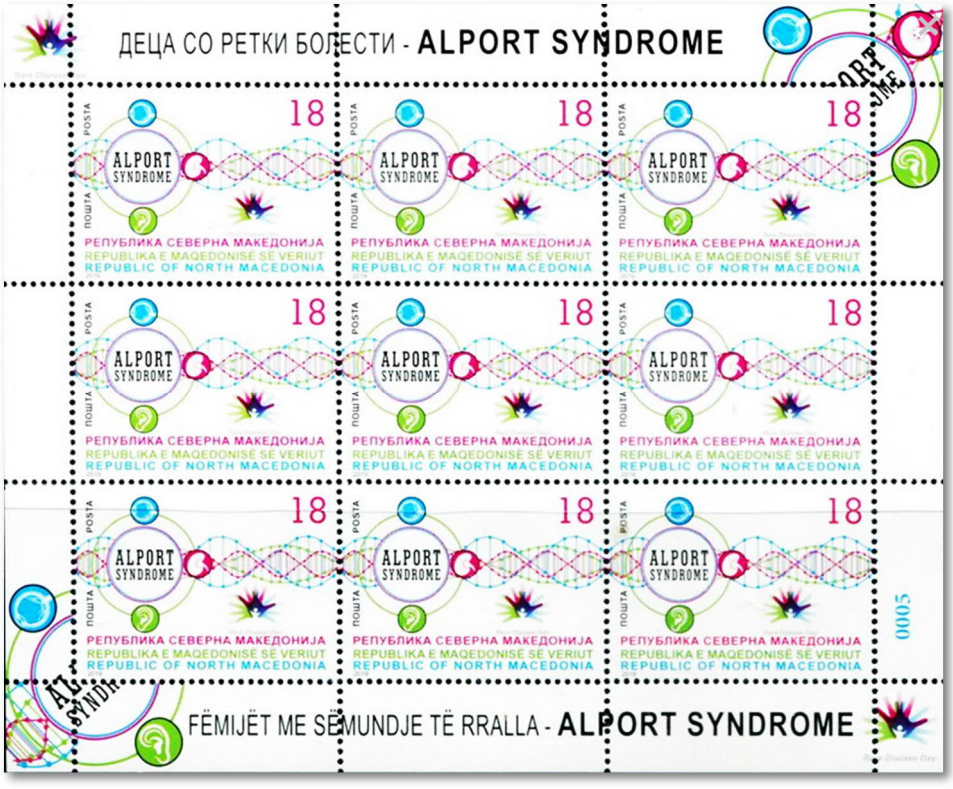
North Macedonia: Stamp series "Rare diseases"

Daniela M. Vogt Weisenhorn

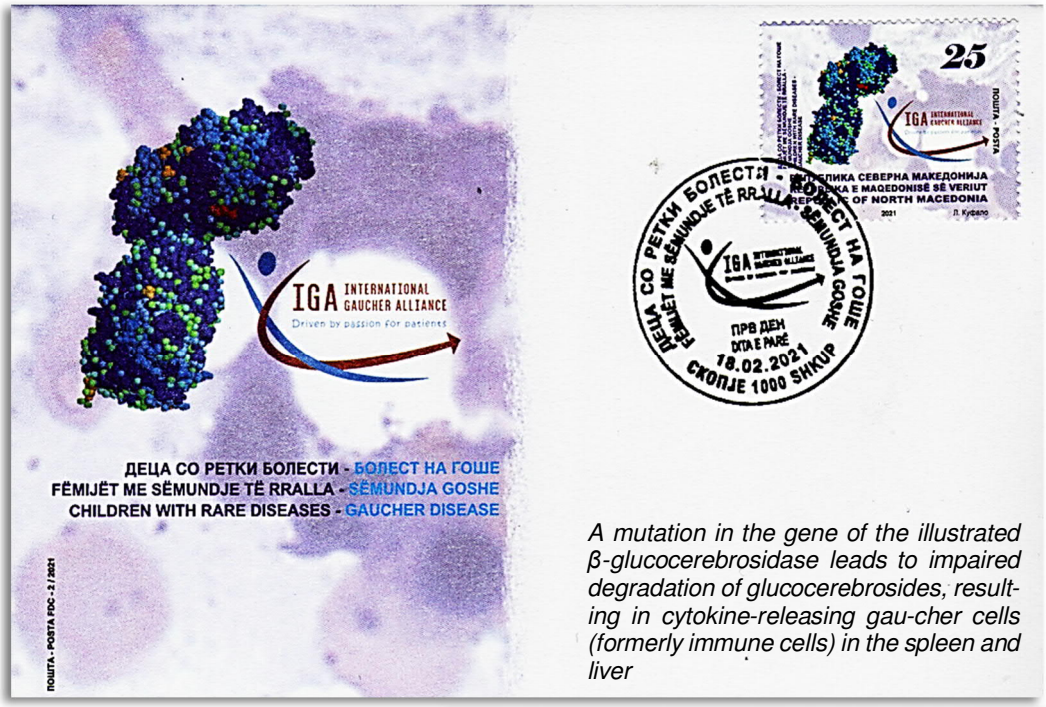
When the son of Gordana Loleska, a postal worker in North Macedonia, was 14 years old, doctors diagnosed him with kidney, vision and hearing problems as Alport syndrome. At first, this diagnosis plunged the whole family into a crisis. However, friends suggested that she get involved in the rare disease initiative.

Four years later, the postal worker has earned a reputation as a committed campaigner for raising public awareness of rare diseases. Much of her success is due to actions that cannot be overlooked.

This included the idea of initiating such a stamp after realizing that there was no stamp on rare diseases anywhere in the world. In no less than two years, she managed to convince the North Macedonian postal authorities that this was a good idea. And so this impressive, globally unique series of 6 stamps was created.



On 10 April 2017, the colorful stamp with the logo of the "Rare Diseases Initiative" appeared as the first in the series. Of these, 6003 were printed. The stamp to raise awareness of her son's disease was then issued on 10 June 2019. Alport syndrome is a rare, inherited kidney disease that can lead to kidney failure. It mainly affects men (80%) and has various genetic causes; mutations in collagen are known. Symptoms include blood in the urine, protein in the urine, sensorineural hearing loss (50%), eye changes (10%) and



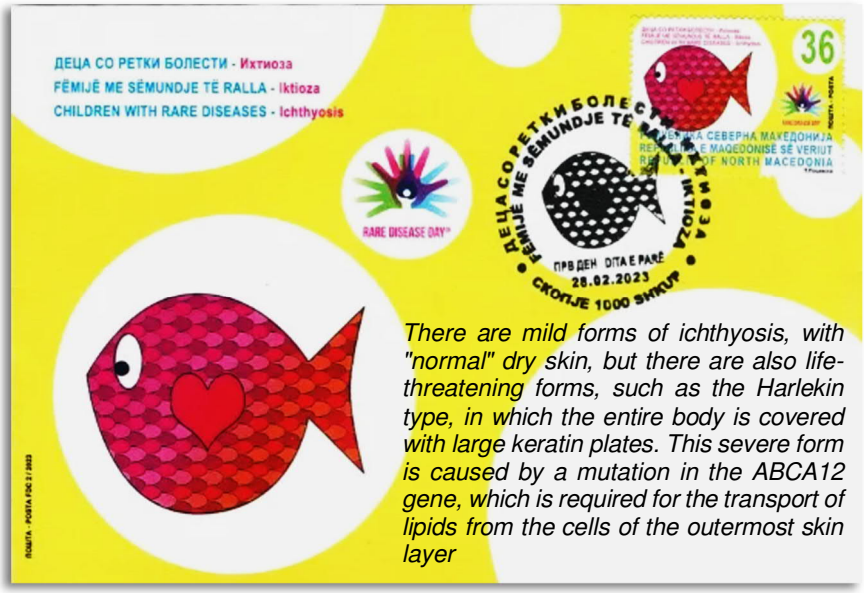
A mutation in the gene of the illustrated β-glucocerebrosidase leads to impaired degradation of glucocerebrosides, resulting in cytokine-releasing gau-cher cells (formerly immune cells) in the spleen and liver

progressive renal failure. Treatment is aimed at slowing down the progression of kidney disease. ACE inhibitors can help, as can early blood pressure control. Dialysis and kidney transplantation are final options.

On February 18, 2021, a stamp was issued with Gaucher syndrome as its theme. The disease (named after the first person to describe it, Philippe Gaucher) is a lysosomal storage disease. Symptoms include splenomegaly, liver enlargement, bleeding disorders and bone disease. Treatment options include enzyme replacement and substrate reduction therapy.

This disease is characterized by mutations in a gene that codes for a protein called Collagen VII. Collagen VII connects the outer and middle layers of the skin with each other

On 28.2.2022 - Rare Disease Day - a stamp was issued to raise awareness of a serious skin disease: Epidermolysis bullosa. Any slight shearing of the skin causes blisters that develop into very painful open wounds. As the skin of affected children is as sensitive as a butterfly's wing, they are called butterfly children. Until now, the only treatment available was wound care. In the last decade, gentherapies have been successfully used in this case - also by applying an ointment.



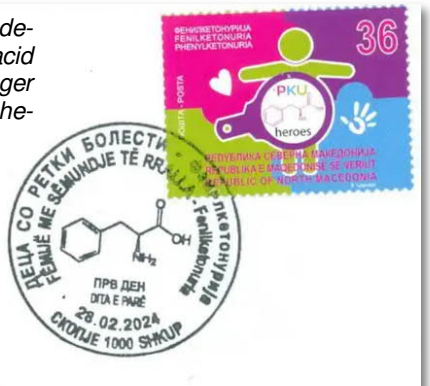
There are mild forms of ichthyosis, with "normal" dry skin, but there are also life-threatening forms, such as the Harlekin type, in which the entire body is covered with large keratin plates. This severe form is caused by a mutation in the ABCA12 gene, which is required for the transport of lipids from the cells of the outermost skin layer

On 28.2.2023 - another Rare Disease Day - ichthyosis was depicted on the stamp. Ichthyosis is a family of genetic skin diseases characterized by dry, thickened, scaly skin. In the first years of life, constant supportive care is required because the extreme scaling leads to dehydration, infections and restricted breathing. Treatment includes

moisturizer, antibiotics and retinoids. About half of those affected by severe ichthyosis die within the first few months.

On leap day in 2024 (29.2.2024), a stamp was issued with the theme of phenylketonuria. If the disease is not treated, it leads to severe brain damage and a progressive delay in psychomotor development, which is noticeable from around the age of 3 months. In addition, there is an increase in spasticity, as well as aggressive, autistic and psychotic behavioral abnormalities. Due to the disruption of melanin synthesis, the disease is also associated with blond hair and blue eyes. A low-phenylalanine diet from infancy onwards can completely prevent the symptoms.

The enzyme phenylalanine hydroxylase, which is defective in phenylkentonuria, converts the amino acid phenylalanine to tyrosine. Because it can no longer convert or break down phenylalanine properly, phenylalanine accumulates in the body....



.... and is alternatively converted to phenylpyruvate (phenylpyruvic acid). The phenylpyruvate accumulates in the blood and reaches the brain, where it causes developmental disorders

I hope that another stamp will be released next year, even though it was originally said that the series would only focus on 5 diseases. With 6000-8000 rare diseases, there is still work to be done. Perhaps in future other countries will also issue stamps on this topic on February 29 - a rare day for rare diseases. It would be nice for the many patients to receive this kind of attention.

Sources: Wikipedia.org, https://med.stanford.edu/news/all-news/2022/03/gene-therapy-epidermolysis-bullosa.html; https://flexikon.doccheck.com/de/Liste_der_seltenen_Krankheiten; orphanet.com

Lazy excuses - the home-brewing syndrome

Clemens M. Brandstetter

ouis Pasteur finally proved that alcoholic fermentation is due to the activity of yeast fungi. Even without this knowledge, alcohol has been produced by fermentation for centuries. For the production of schnapps, kava or beer (France 1973, Netherlands 2011, Luxembourg 2013; Wallis and Futuna 1997; Armenia 2002)

If excuses were to rot, there would be gases due to the decomposition of organic substances in the absence of air. These gases spread an unpleasant smell of rotten eggs, which is due to hydrogen sulphide (H_2S). The term "rotten excuses" is intended to make us understand that an argument is not valid and "stinks to high heaven", so to speak. A similar process takes place during alcoholic fermentation: carbohydrates, usually sugar compounds, are broken down into ethanol and carbon dioxide. Ethanol is an alcohol, a drug that can lead to sensory impairment if consumed excessively by humans. This alcoholic fermentation can also be artificially induced: this typical process takes place in beer brewing, where yeast fungi are used to initiate alcoholic fermentation.

In homebrewing syndrome, you make your own alcohol: from yeasts inside your gut - similar to how you brew alcoholic beer using yeasts. However, our body does not brew "pure" beer - like breweries, but simply alcohol - C_2H_5OH (Germany 2013; South Korea 2019; Perfin Brewery Hatt; Japan 1948 (with chem. formula for alcohol))

There are also people in whose bodies a similar process takes place: this rare disease is called “Eigenbrauer syn-drome”. When we digest the food we have eaten, small amounts of by-products, including alcohol, are also produced. If the human microbiome is disturbed, this can lead to incorrect batches during food preparation. Eating carbohydrates and excessive use of antibiotics can trigger the production of alcohol. Recurrent intoxication with alcohol produced in the body is possible; patients may also exceed the legal limits for driving or operating machinery, which can lead to legal and social problems. The diagnosis of the disease can usually only be made on the basis of information provided by the patient. Determining the alcohol content of the breath at certain intervals can confirm the diagnosis. Antimicrobial therapy and a change in diet can reduce or even eliminate the effects of the disease. Whether the administration of special microorganisms can trigger a health benefit has not yet been clarified.

Back to the lame excuses. Pharmaceutical companies are accused of not researching rare diseases and not considering the development of drugs due to a lack of profit opportunities. It must be remembered that most pharmaceutical companies are simply listed companies - if they were to act against the primacy of profitability, they would have the stock exchange regulator and an army of lawyers with claims for damages on their hands. In order to test a drug on humans, extremely costly preliminary tests, scientific research etc. are necessary; ethics committees have to give their OK and much more - for example, BioNTec is pursuing an approach with its new technology that may make it possible to cure pancreatic or colon cancer (which has so far usually been fatal). The approach has already worked well with COVID (albeit not perfectly; it is a new technology). In the first phase (small sample) of testing this new anti-cancer therapy, there were initial outstanding results (most untreated people died after 2 years - in the sample, more than half were still alive after 2 years, which is incredible progress) - but there are still 2 more phases to go through with increasingly difficult and larger studies before a decision is made in the context of an economic feasibility study as to whether the statutory health insurance companies (can) pay for it at all. Even that is not certain beforehand. And we're talking about billions of euros that such studies cost. Where is a pharmaceutical company supposed to get the money for rare diseases, let alone the people?

Pharmaceutical companies are often listed companies that have to pay attention to profits (Netherlands 2008)

Stamps issued on the subject of alcohol in road traffic: people who suffer from home-brewing syndrome are often unaware that they are driving above the legal blood alcohol limit

Brazil 1995; Denmark 1990; Germany 1971; Israel 1997; Germany 1982; Tonga 1991; Portugal 1982; Spain 2009; Botswana1978; Australia 1990; France 1981; Turkey 1987; South Africa 2004; Vietnam 2020; Portugal 1978; Uruguay, Chile 1996; Guyana (you shouldn't drink and drive in rail transport either) 1988

This is despite studies suggesting that the cost of drug development for rare diseases could be around half that of drugs for common diseases. This may be explained by the fact that the nature of rare disease trials is that they involve fewer participants, are less likely to be randomized or double-blinded, and assess disease response rather than overall survival. All made possible by new regulations on drug approval for rare diseases. However, as a result, many treatments have only been approved on the basis of limited data and surrogate measures, as not only is there little clinical research available, but also relatively little basic medical research on rare diseases, leading to a limited clinical understanding of the disease processes on which new treatments could be based. So everything is completely different from what most people can imagine [quoted from Michael Dobe's mail, April 2024].

Sources: Eigenbrauer-Syndrom - DocCheck Flexikon ; Schlander, M., Hernandez-Villafuerte, K., Cheng, CY. et al. How Much Does It Cost to Research and Develop a New Drug? A Systematic Review and Assessment. PharmacoEconomics 39, 1243–1269 (2021).