Top of Form



Lifestyle

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So rare it is hard to find a cure for disease, APBD Revealed

Only 157 people worldwide, 33 of them in Israel, blue diagnosed to date. Orly Both is one of them. During drug development companies hold due to lack of feasibility, she managed to find a cure

Tags:

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Orly Both. Photo: Ariel Besor

It all began exactly two years ago an innocent skiing Kitzbühel, Austria. Orly Both, organizes conferences and his wife appreciated the Uri Shani, head of the Office of former Prime Minister Ariel Sharon, expertly slid down the icy mountain Flgnstiin, overtaking other skiers on the way down and comes across easily foreclosures. Suddenly she felt a slight weakness in her legs. She tried to slow down the access speed, but stretched out flat on the snow and rolled a few meters down the slope while she was unconscious. Seconds later she opened her eyes and looked at dozens of terrified visitors around her.

"I heard everything they said, but could not get his mouth," recalls both at the same event, after which her life changed when it became clear that She carries an orphan disease. "I wanted to highlight people who are hurt my leg, but the hand did not move. I was sure it was a stroke, but after a few minutes back to my speech. In tests at the hospital it turned out that I got a stroke and I had no breaks in the body, but the weakness did not pass. When I tried to get out of bed, I curled up and fell to the floor. There was no doubt that the fatigue and within two or three days I will repeat myself. I went from the hospital even business conference in Munich when I stumble and relies on friends. "

Monday was certain that shortly after returning to Israel is able to resume sports activities consecutive day. But by this fall has not passed peacefully, but the opposite. She opened a door on a hazardous journey that ended last year's medical diagnosis is not simple. Both suffers from APBD (Adult Polyglucosan Body Disease), a recessive genetic disability that affects motor function initially expressed weakly and legs. The disease, which is characteristic only people from Ashkenazi, actually stems from a defective enzyme is not able to remove the body substance called Foliglokozn, and it accumulates in cells as a poison. The APBD is not fatal and rushes only after age 40, but affects the quality of life and can also spread to the hands or brain and cause cognitive decline rarely.

Given the exceptional disease that is registered in Israel, only 33 patients, some of whom no longer alive because of illness in old age. Worldwide sell only 157 patients. The definition of an orphan disease APBD then, is an understatement. Apparently so rare has made the search for a cure nearly impossible task, partly because pharmaceutical companies working for financial reasons and have no interest in finding a solution to such a small group.

For both the disease is not a difficult but challenging. She turned to renowned experts, the APBD organization and pursues US scientists studying the disease. But apparently it's right under your nose, and entirely the product of Israeli creativity. A meeting between the only specialist in the country to APBD, Professor Alexander Losossna, and a group of Israeli start-up called computational Fftikom, led to finding protein that can repair the damaged structure of the enzyme and stop the symptoms of the disease.

**Orly, so why wait?** "Now are we have two options. The first formula corrects found Fftikom will lead research on animals and humans, which would take several years. The second option and the quickest being tested now is computerized search for cures exist on the market, in the hope that one or two of them will be able to deal with the enzyme defective . If a solution is found this way, the medical history was made here and in two months the APBD patients can start treatment. "

**Sports patient**

Orly Both grew up in Tel Aviv as a child athlete. She won the school championships in the high jump and gymnastics on the uneven parallel bars, and won the Israeli championship shot put. By the time the army draft, is considered to be tested military band, but eventually chose to serve as a sports instructor in the Armored Corps. Upon her release she worked both for the VIP El Al, while enrolled in mathematics and computers at Tel Aviv University, converted after three years studying a degree in Business Administration .

Uri, husband and father of three daughters, she met about 40 years ago, when the two spent together in the resort village of Rafi Nelson Sinai. They went diving together and been together ever since. Two decades ago, two retired airline, and has established two companies the Israeli Opera Center in Jerusalem. A year later entered the field of conference organization, which is engaged in to date, and took part in producing the Jerusalem Business Conference at the end of October 1995.

"This conference will never forget," she said of the beginning of the way. "It was exactly a week before the assassination of Yitzhak Rabin and out was a stormy demonstration. I have no idea why, but I had a feeling that someone hurt him, and I was a terrible shock. At the end of the ceremony of the conference, Rabin sought to examine transportation projects, ignoring the hustle and bustle around. All I wanted was him to go home to avoid any disaster. "

After the conference set up two partner Ruth Koren company conference, Oscar Poor me, engaged in organizing and producing conferences Israeli and international business, especially in high-tech, such as the Innovation Festival, DLD. Among their clients are the Government of Israel, the EU, municipalities Jerusalem and Tel Aviv, New York City Council, Israeli banks and many private companies in Israel and around the world.

Despite intensive work and cross-border, did not give a second one hobby - extreme sports practice. Every winter is staying in one of the well-known ski resorts in France, Austria or Switzerland, and summer skiing snow converts the water. At the same time she loves rough water rafting, and never refused diving into the deep, passionate dances and intense workout in the gym.

In this context becomes even more difficult illness. In retrospect, appeared weak in her legs ten years ago, but two bouncy ignored. "Suddenly I found it hard to jump on stage and high-rise stairs had to hold handrail," she says. "I was not worried that the difficulty was minor and did not hurt me nothing. Sometimes after a long session legs were going numb, so I got up, shook them and moved on. Just before it Skiing in Austria, I felt tired a little walk, but I laughed to myself that I was probably starting to get old."

**Immediately after the crash skiing understanding of your body that something is not right?** "Even then nothing bothered me. Anyone asking why my walking was strange, I answered with some pride that this is a ski accident. A week after returning to Israel, I went to the funeral where I met Prof. Gabi Barabash, the director of Ichilov Hospital. He noticed that I was rocking a little and rejected the excuse of the ski. He asked me to urgently be examined by a neurologist. "

**Shall we?** "Definately not. My motto is to find out the problem and then find ways to deal with it. "

But at no time did not believe the two that Tddh an impasse. First meeting with the neurologist Prof. Nir Giladi, who sent her countless tests. The negative results is received with mixed feelings. On the one hand, ruled out a dangerous disease, yet the mystery of her medical condition has only increased.

"At first I assumed that I had a brain tumor," says two and a sly smile stretched across her lips. "The tests showed white spots on the cortex, but no one knew what they were, and only today we know that one of the symptoms of the APBD. They examined me race brain, nerve conduction, studied chromosomes, and the possibility was raised that I was suffering from ALS or MS, which are degenerative diseases. Even searched in vain for signs of spinal cord injury or Parkinson's disease. The first year I passed all the tests available worldwide, without receiving a diagnosis of some sort. I know there are lots Mistakes about my disease, that is reminiscent of MS, and I have no doubt that there are people among us who have been diagnosed with APBD incorrectly ".
 **What was the most difficult moment at this point?** "Uncertainty. I was living in a black hole. Although I had no fractures or bouts of depression, but there were a lot of questions why walking is better. "

**Father Ashkenazi mausoleum**

Duck Walk lightly swinging of the two, which may cause slight embarrassment to the average person is currently producing only mafia smiles and declarations of love. "It's true that walking cute?" She asks doubt provider prescribes. "In a conference abroad, I introduce myself my name and immediately adds his walk my duck stems from a rare disease. I learned to love it and it does not bother me. I do not need to use a cane or assistance, or have a moment of instability, so I keep a wall or railing. "

**As an avowed fan of reasons, how do you manage to dance?** "I'm not raging as before, but still waving one hand in the air, the other holding someone or something."

**I put on ski and balance.** "Why? I go every year, but not sliding anymore. There's no chance I'll pass on the après ski festivities, hot tub and sauna in the hotel. I believe that I can surf again in the future."

Hard not to be swept away by the bursting optimism and bounding energy of both. She speaks in a row, so check with the messages on the phone, answer e-mails and mostly laughing at herself. Anyone looking for another melancholy or resignation invited to look for another patient. "The disease will not control me," she declares. "I am a practical person and look at everything with cold eyes. At no point will not go into a state of helplessness, because I know deep down that this is a temporary situation."

**How do eventually discovered the APBD you?** "Prof. Giladi overheard talk by Prof. Losossna and sent me to him. I reported to him just a year ago and the problem has heard my first time there. Prof. Losossna sent me for review of the enzyme problem, and two months later it turned out he was right. My initial reaction was happy, at last I know what I have. I immediately asked how to fight, and I realized that the road is still long. "

**What do you know now about the disease?** "All 157 patients with APBD are descendants of the same father Ashkenazi gene ancestor with whiplash. Many of whom live in Israel, and the rest scattered in the United States, Canada, France and Poland. I registered in the country as number 32, and last year also joined a number of patient 33. Then I got sick indeed an orphan, but I made a huge family headed by the great-great-great grandfather one. "

**You know the "family" your remote?** "Through Professor Losossna I referred the request to the Israeli hospital, but only seven agreed to meet and some are already sitting in wheelchairs. We call ourselves' octet secret. Light a Candle met last Tuesday Hanukkah holiday of miracles in the hope that a miracle will also. I have heard terrible stories about people who have had numerous surgeries and faced untold suffering due to wrong diagnosis. It is important that people recognize the APBD and weakness in the legs, and anyone who has doubts about his illness, examined immediately. "

The drug went on the road to finding an accidental meeting between Prof. Losossna and Dr. Amit Michaeli, specializing in computational and medicinal chemistry partner Bfftikom, which is also owned by the Hebrew University. Michaeli is a special algorithm developed by the starting of peptides that bind proteins in the human body, in order to fix them.



The connection between the two experts led a fascinating experience to find a solution for the disease so far known only to Professor Losossna. "We have the patient's protein structure of Orly, and the computer knows the proper protein structure," Michaeli trying to explain words that are not scientific. "The computer is running the algorithm, and a three-dimensional see how different peptides are trying to connect to the problematic protein to which it makes it healthier appropriate".

**What made ​​the startling achievement?** "We took the formula of plant protein, known as Kamchatka due to his letters in a foreign language, and synthesized a white powder which ingredients actually the drug. Meanwhile powder made with first hand experience in the lab, by researcher Dr. Light Hadassah hospital window. He put the Kamchatka cells containing defective proteins hospital and was able to return to activity. "

**Sounds great, how do we go from here?** "There is a long and a short way. The long includes animal testing, testing the drug on humans, certification of the Food and Drug Administration and the like, which would take at least seven years. The shortest way we look at now, and the algorithm scans all formulas drugs approved exist in the world, to find a chemical fingerprint of a protein similar to that facility. "

In terms of the two, this is a real option for the treatment of APBD. "There are cases in which it turned out one drug is also suitable for other problems," she says hopefully. "Take for example the aspirin for headaches today also used blood thinner. If an alternative solution is found, even temporarily, will likely be able to stop the symptoms of the APBD up Designated drug approval. "

**All money**

The race for the right cure for the disease as an orphan in the world was taking shape nor the expertise Lnoirogntikh, Professor Alexander Losossna Hadassah Hospital.
"I was just in the right place at the right time," he says modestly. "I learned about the APBD in 1990, when within a few months I received two patients with the same symptoms. At first I thought it was one of the neurodegenerative diseases, because both had difficulty walking, weakness in the legs and incontinence. Only after many tests I found their precipitation Foliglokozn, resulting from a defect in an enzyme responsible for the construction standards of sugar in the human body. Hfoliglokozn does not fall apart for them, it sinks into the nerves and prevents them from acting. I decided to research the subject and learned that the tiny bodies of Hfoliglokozn were first discovered in the 80s in Canada, but I did not know Nothing on the APBD, because they did not believe that there are genetic diseases that bursts in old age. Nearly ten years I studied the subject until I found the mutation of the gene, and at the end of the 90s were more people carrying the disease. Together with other researchers, we published a medical article on the subject, and it is thanks to Hadassah measures that saved us. "

**There is a chance for a cure soon came to APBD?** "Finding a cure hen will get 50 years in the human body is a complex task and almost impossible, but I'm an optimist. In my opinion, there is a chance to prevent further deterioration with existing drugs, if found. Only then we will be able to access the second goal is to cleanse the body of accumulated sick Mhfoliglokozn ".

Until it becomes clear whether Israel's original initiative was able to draw the benefits of the technology of the 21st century to almost the limit, trying to muster enthusiasm for both the American Association for the APBD patients. But the connection between the Israeli compatible Energizer bunny lovers and Americans bureaucracy creates funny situations relating Meanwhile Sabra chutzpah.

Overseas organization established over a decade ago at the recommendation of Prof. Losossna, in order to raise funds from around the world and deliver grants to researchers of the disease. The American APBD has incorporated dozens of researchers in Canada and Chicago, and their laboratory mice will be used in a few months to examine the Israeli medical formula discovered Bfftikom.

"I do not think I missed any research company in the country and the world," laughs two again. "I learned about my illness and diseases orphans at all, and I discovered how hard it is to find their medication. Everything revolves around money, and a pharmaceutical company does not want to take on a subject that is not economic. I would be happy if the State of Israel and the Ministry of Health had allocated minimal budget for research of diseases orphans in general, that all of us together counters tens of thousands of patients. It's frustrating, because we are basically walking around with a sword over your head and people to calculate their end backwards. In the case of the APBD, octet How much money you can contribute to the development of the drug, because it is our quality of life. "

**Is the organization in the United States helps you financially?** "They announced their intention to participate in the study long and are considering taking part in the short term."

**There has been an experiment in humans, would you take part?** "Even now I am ready to swallow the powder, because in the worst case the material is not going to fight the disease. It will never be known if the project is Professor Losossna, Dr Kahlon and Michael is right unless we try it."

The Health Ministry reported that the orphans are in fact diseases without medication because their development is expensive "rare disease patients are treated in the health basket in spite of the high cost, which is often the million patients a year. The committee which discussed the orphanage diseases, including the definition, treatment and proposed legislation, has completed its work, and its conclusions were submitted to Director General of the Ministry of Health. These recommendations are discussed these days in the work plan for 2016 ".