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Aegerion's representative in Argentina discusses her efforts to raise awareness of Homozygous Familial Hypercholesteremia (HoFH), the rare disease targeted by her company's ultra-orphan drug, and the remaining challenges of diagnosing patients.

What is the current state of affairs for HoFH patients in Argentina?

So far, as lomitapide is not yet approved in Argentina, however, some patients in Argentina have purchased and imported lomitapide on a named patient basis. This mechanism, which was created by a disposition passed in 1995 allows patients to import a product themselves if there is no currently approved therapy, and their physician deems it will help them manage their condition. This has allowed some patients in Argentina to have access to lomitapide, and other unapproved products, on an accelerated basis. As we have helped increase the awareness of HoFH, several physicians have reached out to us when they have a patient with the disease, in one case the patient had already lost a teenage sibling due to what appeared to be a HoFH related medical event, and we have liaised with the head office to help arrange an importation under these rules.

What is the impact of HoFH on the medical system and patient lifespan?

The burden of untreated HoFH is very high, for a very low number of patients. However, the commonly referenced data, which is from studies done in the 1970s, suggests that the incidence of the disease is roughly one in a million, while more recent medical and scientific literature suggest it

could be one in 200 000. Extrapolating from recent data results in an estimated prevalence for HoFH of approximately 6 per one million. Prevalence data continues to evolve, and these calculations could be an under-estimation or over-estimation in the general population.
People with HoFH have a decreased ability to remove LDL cholesterol from their bloodstream, and thus usually have extremely high LDL levels. Thus, common complications of the disease include the problems usually associated with very high LDL cholesterol levels, which may include atherosclerosis, heart attack, stroke, death or other health problems. It is not known whether lomitapide can decrease these problems, or other problems caused by high cholesterol.

How have physicians responded so far to the information regarding HoFH that you have been disseminating?

This is a very new discussion in Argentina and in general the initial response is that they have never seen patients with this disease. However, after we start talking, a number of them have remembered that they had a patient at one point, who despite the usual efforts to help reduce their LDL levels, wouldn’t respond to the treatment. In general, awareness among physicians is quite low, but is now increasing steadily.

What steps is the medical system taking to improve awareness, diagnosis and treatment of HoFH patients?

The system itself isn’t working on anything specific with regards to HoFH treatments, but there are some initiatives including a collaborative initiative between the Argentinian society of cardiology, the Argentinian federation of cardiology, and the ministry of health. This initiative is working to promote cardiovascular health and to decrease the primary risk factors for cardiovascular disease, the primary factors being smoking, diabetes and cholesterol, and I think that this is encouraging the early detection and close monitoring of high cholesterol which would may increase the chance of HoFH diagnosis.

What mission were you given when you joined Aegerion?

My primary task was to learn the market here in Argentina, and to come to understand the environment for Homozygous Familial Hypercholesteremia (HoFH) treatment here. The way in which HoFH is diagnosed and treated varies substantially from country to country because the medical specialty it falls is often different; in the US, there are specialists in lipidology, however no such specialty exists in many countries. In Argentina, there has recently been some evidence that there may be a need for lipidologists and a certification for expertise in lipids was created, however at present, HoFH is often diagnosed by cardiologists, or perhaps the clinician, while in Colombia for

example, it more often falls under endocrinology. Unique to Argentina are nutriologists, who could be described as a specializing in the intersection of gastroenterology, endocrinology, and dietetics, or as nutrition specialists.

Uncovering these unique features was my primary task, along with understanding the difference in the treatment process for a patient living in Buenos Aires city compared to the province, or of course other cities and regions in the country. While the medical system is quite strong in Argentina in terms of coverage and reimbursement, it is still a challenge for many patients to physically access the facilities who can properly diagnose them. As a disease that has been well documented to be under diagnosed and under treated, bringing our treatment to patients relies heavily on understanding the diagnosis process, and working to improve it. There is of course still a lot of uncertainty, as it is unclear exactly how prevalent the disease is here in Argentina relative to other populations, such as in the Netherlands, where it is relatively common and well diagnosed.

What does this diagnosis process usually look like?

The actual process depends quite a lot on where they live, their age, whether or not they have some form of health insurance, and so there isn’t a single, universal diagnosis algorithm. Moreover, the disease can manifest in many different ways, at different stages in life, leading to the patient seeking attention from different specialists. For children, Xanthomas (wart like cutaneous lipid deposits) are typically the first identifiable symptom, and as such their parents may take them to a dermatologist. Other patients with less severe presentations of the disease may go undiagnosed until their early adulthood, when they might have their cholesterol checked for the first time and their GP finds a LDL level of 600. In general, the condition is still diagnosed symptomatically, and often based on the evidence that patients do not respond to traditional anti-cholesterol medications as expected.

There are genetic tests for HoFH, but the Argentinian Society of Lipids current position is that the testing is not required for treatment. Currently, over 1000 different mutations that play a role in HoFH have been characterized, there have not been studies to determine which mutations are most common in the Argentinian population, and there are likely many more that are not currently understood; thus, any genetic tests may be incomprehensive and may have a high false-negative rate. Furthermore, because treatment is on a clinical basis, even a positive result from a genetic test may not affect the prescribed therapy.

Based on your experience, how well is Argentina positioned to manage rare diseases from a policy standpoint?

As you know, there is new legislation now for orphan drugs, but very few products have been registered using these new mechanisms so far. I think it is great that there is recognition that there is a difference in the challenges orphan drug distributors face, and that they are setting the groundwork for a more appropriate registration process and testing regime for these products. Needless to say, it still remains to be seen how well the system functions once this law is fully implemented and used regularly.

What would you like to accomplish for Aegerion and HoFH patients in Argentina over the next five years?

Johann Wolfgang von Goethe once said “you see what you look for, and you look for what you know”, which concisely sums up the critical need for us to increase the level of awareness surrounding this condition. So over the next five years, I would like to see substantive evidence of better awareness of HoFH, and hopefully some indication that the rates of diagnosis and treatment have started to increase as well.

How did you become involved with Aegerion?

I am originally from Uruguay, and studied biology at the Universidad de La Republica in Montevideo before doing graduate studies in genetics. Afterwards, I moved to the United States on a scholarship at Tulane University in New Orleans, but decided I wasn’t interested in pursing a career in basic research any further and started looking for an MBA program near a strong biotech environment. Eventually, I moved to Boston, did an MBA at Boston University, and then started working at Biogen Idec as a product manager. In 2004, I was hired by Genzyme and moved back to Latin America before transferring over to Sanofi after the acquisition. In early 2013 some former Genzyme colleagues who were working at a biopharmaceutical startup called Aegerion contacted me with an offer to join them, and after some time considering, I accepted.

Initially, I didn’t think that I would accept the offer because I was quite comfortable in my position at Sanofi. However, I did agree to have an interview with their team and after getting to know them a bit they seemed like a really great group of people with a great sense of energy and who were determined to make a real impact, and I found myself feeling less excited by my work at Sanofi than I had been before. What really convinced me was that while many companies claim they have global aspirations, Aegerion very clearly was delivering on that ambition by deciding to bring their therapy to patients in need around the world.

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