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Rare Disease Day and Blue Cone Monochromacy – Press Release

Urgency of structural funding for diagnosis and gene therapies

Jupiter, FL - Today is Rare Disease Day, and patient organizations around the world are dedicated to creating visibility and understanding for their disease. We speak today with Dr. Renata Sarno, President of the BCM Families Foundation, dedicated to a rare eye disease, Blue Cone Monochromacy.

Dr. Sarno, what disease is Blue Cone Monochromacy?
Blue Cone Monochromacy (BCM) is a rare disease of the retina that occurs in 1 over 100,000 newborns and causes low visual acuity, poor color discrimination, aversion to light, and nystagmus from the first months and throughout life. It is a disease that affects an average of 4 to 5 people in each affected family, exclusively male because it is transmitted via the mother on her X chromosome. The children have low vision problems which compromise their school and personal life.

What do patient organizations want?
We need diagnosis to be readily available for all families who suspect their child has this disease, with targeted screenings and for ophthalmologists and pediatricians to be able to diagnose Inherited Retinal Diseases. But, above all, that gene therapies are also financed with public support and lead to genetic drugs available to patients at not exorbitant but affordable prices.

What is the biggest obstacle today in bringing gene therapies for these eye diseases to the market?
Currently at least 270 genes are known to be associated with Inherited Retinal Diseases and many more genetic causative mutations are known. Today the obstacle to finding a genetic cure for all these mutations is that the process used to develop each one is very expensive and slow. In the last 10 years only 1 or 2 drugs have come on to the market but at exorbitant prices, exceeding $750,000. We urgently need to find models for the efficient development of genetic treatments, which can be used in parallel on multiple genes and on multiple mutations. It is not possible to leave this investment in the hands of a few biotechnological companies at the mercy of the fluctuations of the financial market, but a suitable and structured development model is needed.

How long have you been involved in the BCM path toward a cure?
I have been involved with finding a cure since 2009, after receiving a diagnosis of BCM for my son. In all those years I have dedicated myself to bringing together a community of isolated rare patients scattered all over the world and to consolidating their medical data on to a patient registry and in some research centers. Every year many people contact me, even with other eye diseases, or who after so many years are unable to give a name to their disease, asking me where to access the DNA test, if there is any news on gene therapy, or help to develop local resources for children going to school. We run a newsletter and information webpages on visual aids, expert centers, and family meetings. Each contact is a new story. A few weeks ago, Ellen from Sweden called me and told me about her children. She has 3 young children, one of them diagnosed with PDE6H Achromatopsia, the other two also show the same symptoms. I immediately thought of my son with BCM, of the efforts we are concentrating to find a gene therapy, and of the hard work from 2009 to today, involving research centers and carrying out clinical and pre-clinical projects which still do not see a concrete result. But this mother has three children with a disease similar (although rarer) than ours -- What will become of her? What can I tell her? And in this period in which biotech companies are halting research on drugs for rare diseases due to lack of funds, even after years of involving patient associations, I realize that we are going the wrong way. I just know that whichever path we take, we will take Ellen with us, the rarest amongst the rare, because this is the only way out for everyone, this is how it works among us humans: we cannot save ourselves alone!

BCM Families Foundation is a non-profit association established and led by patients and their families, with a mission to find a cure for Blue Cone Monochromacy. It supports scientific research and collaborates with numerous research institutes in the USA and Europe, with biotechnology companies and with other patient associations.
Find more at www.blueconemonochromacy.org

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