

Chiesi at EPNS 2023 to confirm its commitment on alpha-mannosidosis with the projection of the movie “Rare Land” and the presentation of 4 posters

Published On: 2023-06-23 00:00

Category: #WeACT



- “*Rare Land*” is a movie by Eftimiz Hatzis, which gives the audience a front-row seat to understand the difficult experience of a family facing a rare disease, such as alpha-mannosidosis.
- Challenges in the early diagnosis of rare and ultra-rare diseases and opportunities to increasingly support patients (also via new means of communication such as cinema) were among the topics discussed at the event.
- Chiesi is a player attentive to raise awareness and bring significant changes in the rare disease field: 4 new posters illustrate recent results related to the disease and treatment of patients with alpha-mannosidosis.

Prague, June 22, 2023 – Chiesi participated in the 15th edition of the **European Congress of Paediatric Neurologists (EPNS)** where the company organized a screening of 'Rare Land', a film on alpha-mannosidosis realized by the Greek director Efthimis Hatzis with the unconditional support of **Chiesi Global Rare Diseases** - also presented recently at the Cannes Film Festival, in the Marchè du Film/Short Film Corner section and at the Greek Film Centre Pavillon - and presented a **series of posters on the treatment of this disease**. The screening of the film was followed by a discussion with **Ass. Prof. Martin Magner**, MD, PhD, Department of Paediatrics and Hereditary Metabolic Disorders of the General University Hospital of Prague, and **Prof. Dimitrios Zafeiriou**, Professor of Paediatric Neurology and Developmental Paediatrics, 1st Department of Paediatrics at the Hippokratio General Hospital of the Aristotle University of Thessaloniki in Greece, **the first physician to discover a case of alpha-mannosidosis in Greece**, which was moderated by the Italian journalist Federica Gentile.

"Rare Land" tells the story of Alexandros, a child suffering from the ultra-rare disease **alpha-mannosidosis** and provides a **unique insight into the emotional journey he and his family go through** from the onset of the first symptoms to the final diagnosis, amid feelings of fear, bewilderment, doubt, and hope.

"Films like 'Rare Land' are important because they allow us to realistically show what happens in the daily lives of patients living with ultra-rare diseases and of their families, shedding light on diseases that often risk being forgotten" commented **Prof. Dimitrios Zafeiriou, Professor of Paediatric Neurology and Developmental Paediatrics, 1st Department of Paediatrics at the Hippokratio General Hospital of the Aristotle University of Thessaloniki in Greece.** – *"Tackling topics like this through a non-traditional communication medium for this sector, such as cinema, allows the audience to be completely immersed in the stories of patients and their caregivers and offers an intimate and impactful perspective on a delicate and still unknown issue."*

Alpha-mannosidosis is an ultra-rare disease, **affecting approximately 1 in 500,000 to 1,000,000 children born worldwide** and is caused by a mutation in the MAN2B1 (LAMAN) gene responsible for the production of the enzyme lysosomal alpha-mannosidase, which is absent or sub-optimally expressed in affected individuals. This deficiency in enzyme activity results in the **inability of cells to break down complex sugars**, the oligosaccharides, which then accumulate in a progressive and damaging manner throughout the body. The disease, which can vary in severity, can manifest itself at a young age through various symptoms, including **recurrent respiratory tract and ear infections, hearing loss, distinctive facies, muscle weakness, skeletal and joint anomalies, visual and cognitive abnormalities.**

Improving the diagnosis journey for this disease, which is still often long and difficult, and **ensuring early diagnosis** are crucial to truly make a difference for patients, in many cases.

"Pediatric neurologists play a crucial role and are key figures in correctly identifying patients' symptoms in a timely manner and in effectively managing their care" explained **Ass. Prof. Martin Magner, MD, PhD, Department of Paediatrics and Hereditary Metabolic Disorders of the General University Hospital of Prague.** *"Especially for this kind of disease, where early diagnosis has a direct effect on the patients' future prognosis, pediatric neurologists, along with the broader multidisciplinary team, is a key point of reference that should directly prescribe the most appropriate treatment, where possible, or refer the patient to a specialized metabolic center".*

Chiesi is committed to **improving the lives of people with rare and ultra-rare diseases and their caregivers.** As part of this commitment, the company enthusiastically supported this project to **raise awareness on the issue** and offer a realistic insight into the journey faced by those who receive this diagnosis.

*“At Chiesi Global Rare Diseases we constantly strive to support patients of the rare disease community by listening to their needs and helping improve the patient journey”, said **Enrico Piccinini, Head of Europe, and Emerging Markets Rare Diseases at Chiesi Group**. “We are proud to participate in such a prestigious Congress as the EPNS, sharing with the medical community the touching testimony of 'Rare Land', which represents the real experience of many families facing a rare disease diagnosis. As a company, we are inspired by stories like this to find innovative solutions to address unmet needs and to spread awareness about rare diseases”.*

At the **EPNS** congress, Chiesi underlined its commitment in this area also through the **presentation of 4 posters** illustrating recent results of **clinical trials related to the disease and treatment of patients** with alpha-mannosidosis with velmanase alfa, an enzyme replacement therapy developed by Chiesi as a recombinant form of human alpha-mannosidase, intended to supply or supplement the natural enzyme, which is deficient in patients.

Find out more about “Rare Land” and the issues addressed in the film at <https://wayfinding.chiesi.com/premiere-rareland/>

About alpha-mannosidosis

Alpha-mannosidosis (?-mannosidosis) is an ultra-rare autosomal recessive disorder caused by a mutation in the MAN2B1 (LAMAN) gene which encodes for an enzyme called lysosomal alpha-mannosidase that is lacking or sub-optimally expressed in affected individuals. The reduced activity of the enzyme results in an intracellular (lysosomal) accumulation of oligosaccharides (a complex of 2–10 simple sugars), which is toxic to cells and organs. Oligosaccharides can potentially accumulate throughout the body and affect multiple systems.^[1]

The disease affects approximately 1 in 500,000 - 1,000,000 babies born worldwide.^[2]

According to autosomal recessive inheritance, offspring of carrier parents have a 25% risk of being affected whereas 50% will be non-symptomatic carriers.

There are 3 forms of ?-mannosidosis:

1. Type 1: a clinically mild form recognized after 10 years of age. This form has a very slow disease progression and does not present skeletal abnormalities
2. Type 2: a moderate form recognized before 10 years of age. This form has as well a slow disease progression and presents skeletal abnormalities with a subsequent development of ataxia occurring from 20-30 years of age.
3. Type 3: a severe form that is immediately recognized and presents itself with

skeletal abnormalities and obvious disease progression. This form results in early death from CNS involvement or myopathy^[3]

Further information on the disease is available at the following site:

<https://www.alphamannosidosis.com/en/for-patients-caregivers/>

About Chiesi Global Rare Diseases

Chiesi Global Rare Diseases is a business unit of the Chiesi Group established to deliver innovative therapies and solutions for people affected by rare diseases. As a family business, Chiesi Group strives to create a world where it is common to have therapy for all diseases and acts as a force for good, for society and the planet. The goal of the Global Rare Diseases unit is to ensure equal access so as many people as possible can experience their most fulfilling life. The unit collaborates with the rare disease community around the globe to bring voice to underserved people in the health care system.

For more information visit www.chiesirarediseases.com.

About Chiesi Group

Chiesi is an international, research-focused biopharmaceuticals group that develops and markets innovative therapeutic solutions in respiratory health, rare diseases, and specialty care. The company's mission is to improve people's quality of life and act responsibly towards both the community and the environment.

By changing its legal status to a Benefit Corporation in Italy, the US, and France, Chiesi's commitment to create shared value for society as a whole is legally binding and central to company-wide decision-making. As a certified B Corp since 2019, we're part of a global community of businesses that meet high standards of social and environmental impact. The company aims to reach Net-Zero greenhouse gases (GHG) emissions by 2035.

With over 85 years of experience, Chiesi is headquartered in Parma (Italy), operates in 31 countries, and counts more than 6,500 employees. The Group's research and development centre in Parma works alongside 6 other important R&D hubs in France, the US, Canada, China, the UK, and Sweden.

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[1] *Malm D et al. Orphanet J Rare Dis 2008;3:21*

[2] *Harmatz et al. Molecular Genetics and Metabolism 124 (2018) 152–160*

[3] *Malm D et al. Orphanet J Rare Dis 2008;3:21*