FEBRUARY 28, WORLD RARE DISEASE DAY

Film and literature surrender to rarity

On the big screen and in the novels the characters can be as tall as giants, of short stature or with facial alterations. And it's not about fiction: they suffer from rare diseases. The patients celebrate that their ailments appear in the cinema and in the literature, but they ask for more realism and that their presence stops being exceptional.

Laura Chaparro 28/2/2018 08:00 CEST

The movie 'Wonder', released in 2017 and based on the book of the same name in 2012 by Raquel Palacio, tells the story of a child with Treacher Collins syndrome. Julia Roberts plays the role of her mother and Owen Wilson, her father's. / LionsGateEntertainment

Being born healthy or sick can depend on a genetic mutation. As a reflection of reality, some films and novels have wanted to immerse themselves in these pathologies and portray the life of those who suffer them. On the occasion of the World Day of Rare Diseases celebrated on February 28, we have asked patients' associations how they have seen their illnesses treated in the cinema and in literature. We have also talked with writers and agree: these works make rarity visible and normalize it.

Acromegaly

The giant of the Goya Awards

En Miguel Joaquín Eleizegui, a man with a peculiarity: he came to measure 2.42 meters, lived in the small municipality of Altzo, in Guipuzcoa, Basque Country. The story of the *Giant of Altzo*, as he was nicknamed, is narrated in the film *Handia* (2017), winner of ten Goya Awards, among them, best original script.

'Handia', 2017.

Uno One of the scriptwriters, José María Goenaga, contacted the Spanish Association of People Affected by Acromegaly three years ago. Its president, Raquel Ciriza, tells us that she provided information about the disease when the movie was being documented. According to Ciriza, the scriptwriter told her not to expect to see a reflection of reality since, although the film was based on a real character, it was fiction.

In her opinion, the final result is positive because it shows the suffering of the patient, who in the film receives no treatment to control his illness, and the treatment they give him for being different, turning him into a claim to do business.

The acromegaly of the protagonist of '*Handia*' is produced by an excess of secretion of growth hormone

What does not just agree with reality is that the character began to increase in size past adolescence. Acromegaly is an endocrine disease that is caused by an excess of growth hormone secretion due, in most cases, to the presence of a benign tumor in the pituitary, a gland in the cranial base.

The disease causes enlargement of tissues, organs and extremities, such as hands and feet. "If it happens to you in adulthood, you have acromegaly and you do not have to be taller than normal. However, if the excess of the hormone occurs in childhood or adolescence, gigantism is talked about, as it grows high because the cartilage has not finished closing", explains Ciriza to Sinc. In the case of the Altzo Giant, the disease had to start earlier than the film narrates.

Other mythical acromegaly giants of the big screen were the fearsome 'Shark' of James Bond, played by the actor Richard Kiel, and the loyal Fezzik of *The Princess Bride* (1987), which represented André René Roussimoff, a professional wrestler.

Achondroplasia

The low size is not funny

At the opposite pole are people suffering achondroplasia, a bone disease in which cartilage growth is interrupted and causes shorter limbs and short stature, among other symptoms. Recent films like *Summer 1993* (2017) and *Three Billboards Outside Ebbing, Missouri (*2017) include supporting roles with this rare pathology.

Three Billboards Outside Ebbing, Missouri (2017)

In an article published in *The Guardian*, Eva Squire, who also has the ailment, denounces the discriminatory and comical treatment that is given to the character of *Three Billboards Outside Ebbing, Missouri* played by Peter Dinklage. "In the movies, it is still impossible to find complex, serious characters who face conflicts derived or not from their dwarfism. They appear as extras to put a comical touch or offer a contrast", spokespeople of the Spanish Fundación ALPE Achondroplasia foundation explain to Sinc.

" It is still impossible to find complex, serious characters who face conflicts derived or not from their dwarfism", patients complain

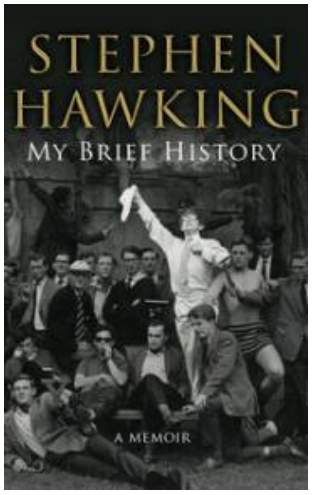
For these patients, the only exception is *Short Cuts* (2003). Also played by Dinklage - well known for his participation in *Game of Thrones* - the film realistically narrates what it is like to live with the disease. "Dinklage is a model for us. We admire him and we value him very much. He has achieved what no actor with achondroplasia or other forms of dwarfism had achieved before: triumph for his quality as an actor", they emphasize.

In *Willow* (1988) or *Simon Birch* (1998) the protagonists also present forms of dwarfism, although not achondroplasia. From ALPE they admit a small evolution, especially in television. "We accomplished that in the *1,2,3 TV Show*, the regular appearance of Tati and Quieti, two people with achondroplasia who acted as unruly children of a man dressed as a woman,

was suspended," they stress.

Amyotrophic Lateral Sclerosis

Beyond Stephen Hawking



ALS - amyotrophic lateral sclerosis_- is another rare disease that has appeared on the big screen. *The theory of everything* (2014) tells the life of Stephen Hawking, the famous physicist who was diagnosed with the neuromuscular disease when he was 21 years old.

Based on the memories of his ex-wife, Jane Hawking, the film does not realistically reflect what life is like for a patient, according to Rosa María Sanz, manager of the Spanish ALS Association."It's even harder and the progression of the disease, in general, much faster," she says.

This disease of the central nervous system is characterized by a progressive degeneration of motor neurons in the cerebral cortex, brainstem and spinal cord. The consequence is a muscle weakness that can progress to paralysis.

In the book *Brief history of my life* (2014) Hawking takes stock of his career and tells how he has faced the ALS. "Why did it have to happen to me? At that time I thought that my life was over and that I would never develop the potential I felt I had. However, now, fifty years later, I can be satisfied with my life," he wrote.

According to Sanz, the scientist is a special case, which does not represent the majority of patients, and the film is not focused on the ALS and its evolution, but on his specific case. "It is necessary to see how a patient is confronted with the acceptance of the disease and how he is living the duels of the successive losses that he is suffering. Nor are family or personal decisions made regarding death and life seen in the movie," she maintains.

Treacher Collins Syndrome

“I know I'm not a normal ten-year-old kid”

In *Wonder* (2017), Julia Roberts and Owen Wilson play the parents of August "Auggie" Pullman, a child with Treacher Collins syndrome. The film is based on the homonymous book written by Raquel Jaramillo Palacio. "I know I'm not a normal ten-year-old," Auggie says in the novel. Both he and the other characters in the play are fictional.

'Wonder', 2017

The Treacher Collins syndrome is a congenital craniofacial malformation characterized mainly by the absence of cheekbones, ears, cleft palate, and digestive and respiratory problems. In the book Auggie talks about the twenty-seven operations practiced with him, something quite realistic. However, according to Marisa Gil, president of the National Treacher Collins Syndrome Association, both the novel and the film soften reality.

In ‘Wonder’ the vision of parents and how they reconcile work and family with life in hospitals is missing

“It is an adaptation that does not reflect the real life of a child with the syndrome, since it is not normal for parents to allow themselves the luxury of stopping their jobs to teach their child, to start school in 5th grade, or much less wear a helmet," she says.

To avoid the looks and whispers of others, Auggie used for several years an astronaut helmet a friend of his sister has given him. In the book the narrator starts being the child but then so are his sister and his friends, which allows knowing their points of view. However, the vision of the parents does not appear.

"We need to see how his parents see it and how they try to reconcile work and family with hospitals," says Gil. What seems positive for the visibility of the disease is that actors such as Julia Roberts and Owen Wilson participate in the film.

Friedreich's ataxia

Patients and writers

Except in the case of Stephen Hawking, the writers or directors of the books and films we have mentioned are not patients, something that changes with *The legacy of Marie Schlau* (2014). It is a collective novel written by 17 authors, almost all women, and most with Friedreich's ataxia. The funds raised with the sale of the book are to research the ailment.

Friedreich's ataxia is a hereditary neurodegenerative disease that produces progressive lesions in the nervous system and causes muscle weakness, speech problems and heart disease, among other symptoms. It was the philologist and patient María Blasco Gamarra who had the idea of this collective work.

"I thought that in different parts of the world there should be people with the same disease and a common goal: investing in biomedical research to find some effective treatment," she says. She contacted the BabelFAmily association, which promoted the project and translated the text altruistically. The authors of the novel come from Spain, Australia, the United States, Mexico, Portugal, the United Kingdom and South Africa. One of them, Nicola

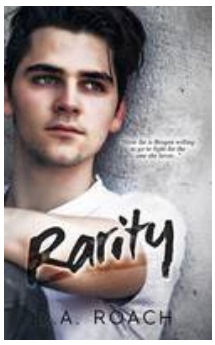
Batty, died before it was published.

The protagonist of the book, the young Marie Schlau, suffered from the disease in the first half of the nineteenth century, before it is diagnosed. Although it is a work of fiction, real characters appear as the neurologist Nikolaus Friedrich, who described the disease in 1863. In the novel, Marie transmits the disease to her great-great-grandson Ron. "I am a prisoner of a genetic code that feeds on my descendants," the young woman laments in the book.

Although sales are not being high, Blasco Gamarra is optimistic because the proceeds fund research projects. "The beneficiaries are not just us, the patients, but all of humanity," she stresses.

Ehlers-Danlos syndrome

The therapy of a mother



After several years without knowing what happened to her son, and once autism and attention deficit and hyperactivity disorder (which her other daughter suffered), had been discarded, a geneticist solved the mystery: the boy suffered from Ehlers-Danlos syndrome of vascular type. "The diagnosis was devastating," recalls Deborah A. Roach.

This rare disease encompasses a group of incurable hereditary disorders characterized by extremely lax joints, very elastic skin where bruises easily form and blood vessels that are easily damaged. Patients who suffer from vascular syndrome, such as Deborah's child, have a very high chance of a rupture of an organ or a major blood vessel.

After turning to her son, talking to other patients and learning everything about the syndrome, Deborah went to a therapist. "I wanted to make sure I was not so caught up in the diagnosis that I forgot to live," she says. As a therapy, she decided to write a fiction book in which the disease appeared: *Rarity* (2015).

The novel is a story of adolescent love in which the protagonists fight against many obstacles, among them, the syndrome. The reception by the patients was very good, although for some of them it was like a jug of cold water. "They know the data - a short life expectancy - but they try to hide that information in the deepest recesses of their minds", explains the writer.

Williams syndrome

Trust patients

Journalist Jennifer Latson remembers very well when it was the first time she heard about the Williams syndrome: in a news story. In it, patients were described as biologically incapable of distrusting. "At first I was surprised to hear that this was considered a disorder because it seemed like something we should celebrate, not cure," he acknowledges.

“Eli is the only teenager I've met who told his mother several times a day: 'I love you mom, you're the best”

Cuando When she investigated the genetic disease, she knew the most serious symptoms that accompany it, such as cardiovascular problems, some type of mental retardation or long facial features. She met Eli D'Angelo, who had the syndrome and his mother Gayle, and followed them closely for three years, when Eli was between twelve and fifteen years old. That period was captured in the book *The boy who loved too much* (2017).

"Eli is the only teenager I've met who told his mother several times a day: 'I love you mom, you're the best,'" says the journalist. The patient associations thanked her for dedicating the book to the syndrome and using a language accessible to the general public, avoiding medical technicalities.

The novel tells the difficult decision of the mother, who must choose between protecting her son from the rest of the world to avoid suffering or give him more freedom, despite the setbacks that arise. A tessitura that is also reflected in other works and well known to the mothers and fathers of any child with a disease. Thanks to film and literature, these families, so different and similar to each other, feel less and less rare.

The film festival of rare diseases

The Disorder Festival. / EricaDerrickson

In this festival there are no losers. Everyone wins, above all, patients. Its founders, Bo Bigelow and Daniel DeFabio, came up with the idea after recording two films about the rare diseases their children suffered. "We wanted to create a festival where we could show our films, select others and bring together an audience that includes activists, researchers, doctors and pharmaceutical companies," summarizes Bigelow to Sinc.

Disorder: TheRareDisease Film Festival was held in Cambridge (USA) last October and brought thirty films together, among them the Spanish *Cuerdas* (*Strings)* (2013), which in 2014 won the Goya Award for best short animation. Other countries represented were Poland, Iran, Canada, the United States and the United Kingdom. The organizers want to reach 7,000 films, as many as rare diseases and to accomplish this goal, they put directors in contact with patients who want to see their disease on the screen.

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