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THE DEBATE ON RARE DISEASES

A LOOK AT MEDIA RESPONSE

ANTONIO M. BAÑÓN HERNÁNDEZ AND JOSEP A. SOLVES ALMELA

Rare diseases (RDs) are those that affect fewer than five people in every 10,000. There are around 7,000 RDs, they are difficult to diagnose and very few have a treatment. This article explores how the media report on the arguments and counter-arguments regarding the access to drugs for these pathologies, with critical discourse analysis for the case of Duchenne muscular dystrophy. We concluded that, in times of crisis, the debate polarises around economic arguments (the price of drugs), reinforced by doubts about their healing effectiveness.

Keywords: critical discourse analysis, rare diseases, means of communication, argumentation, orphan drugs.

RARE DISEASES

Rare diseases (RDs) affect fewer than five people in every 10,000. There are around 7,000 pathologies identified as *rare*, and a high percentage have a genetic origin. They are sometimes very difficult to diagnose, and very few have a treatment. Although the situation has improved in recent years, research is still insufficient, so the cure or improvement horizon is limited. Social unawareness of RDs can sometimes also be noted in healthcare environments. In addition, many of them are also very serious and highly incapacitating, especially when manifested during childhood.

The Study on the Socio-Sanitary Needs of patients with Rare Diseases and their families (ENSERio, Federación Española de Enfermedades Raras, 2009) revealed that 50 % of these people are not satisfied with their health care and that the estimated time to receive a diagnosis is five years. Almost 80 % of interviewees said they felt they had received inadequate attention at some point and 77 % claimed to have been discriminated against on occasion due to their illness.

However, beyond this information, there is barely any research on the social and humanistic aspects of RDs. Although the study of the process of symbolic construction of these pathologies has already started in the media and in social networks (Armayones, Requena, Gómez, Pousada, & Bañón, 2015; Bañón, Fornieles, Solves, & Rius, 2011), as has the study of the impact they have on doctor-patient interactions (Budych, Helms, & Schultz, 2012), a detailed analysis of the political and economic arguments used in the RD debate is still pending. This is the reason behind the focus of this article.

PUBLIC DISCOURSE ON RARE DISEASES

In spite of all of the mentioned above, RDs have found a sort of stable space in the scientific, media, political and institutional, and social agendas in the last fifteen years (Solves, Bañón, & Rius, 2015). The main protagonists, affected people and their families, have made an effort to communicate well, and each of the fundamental groups have differentiated messages. They have tried to convince politicians and decision-makers that RDs should be considered a health priority issue today.

«THERE ARE AROUND 7,000 PATHOLOGIES IDENTIFIED AS "RARE", AND A HIGH PERCENTAGE HAVE A GENETIC ORIGIN»

But in reality, the political and the economic debate have always overlapped. An editorial in *The Lancet* (May 2015) titled *Reducing the cost of rare disease drugs* described the cost of orphan drugs –developed with public incentives because they are

not profitable for pharmaceutical companies – not without certain irony, as an «unwanted side-effect». Similarly, some prestigious scientific journals do not hesitate to publish research headlines such as the following: «Expensive drugs for rare disorders: to treat or not to treat?» (Schlander & Beck, 2009). It is easy to focus on the price of drugs when the economic situation is one of the main concerns of society. However, the association between RD drugs and high prices sometimes generates a metonymy that equals rare diseases and expensive patients, and with that metonymy comes a paradox: the solution (finding a cure) becomes the problem (financing it).

In an interview, the General Secretary of the Spanish Ministry of Health, Social Services and Equality stated: «Some countries have not included the new drugs because they were not cost-effective. We have to accept that the cost of innovation is extraordinarily high, some drugs for rare diseases cost 300,000 euros. The problem is that some of them do not even cure» (Marqués, 2015).

The fragment is very relevant for debate, because of the speaker's position and because of what he said. He referred to approved and commercialised drugs. He directly mentions RDs as an example of expensive diseases, and just as importantly: he says some drugs do not even cure. This approach considers only two categories: being ill or being cured, black and white. No greys. We should debate about deep economic perspectives that also consider ethics and science, such as: should we invest in those drugs if they stop the disease, even if they do not cure it? What do we do when they slow the disease? What if they offer a slight improvement?

Social unawareness of rare diseases can sometimes also be noted in healthcare environments. The picture shows an official poster for the Rare Disease Day, celebrated on the last day of February each year in order to raise awareness of these diseases and their impact on patients' lives.

The authorisation to market an orphan drug does not necessarily involve automatic funding by the administration or insurance companies. In fact, there are important differences among European countries on this matter (Picavet, Annemans, Cleemput, Cassiman, & Simoens, 2012). To secure financing, it is usual to allude to the cost-effectiveness or cost-utility of the drug (Graf & Frank, 2015). In practice, the decision to fund orphan drugs often derives from the combination of «official» factors (therapeutic value, impact on the budget, price and impact in clinical practice) and «unofficial» factors like comparison with other countries, the influence of patients and experts associations, the ascription of the pathology to adults or children, the media's reaction, the innovative nature of the product, the political climate or ethical considerations (Picavet, Cassiman, & Simoens, 2014).

«RARE DISEASES ARE THOSE THAT AFFECT FEWER THAN FIVE PEOPLE IN EVERY 10,000»

The objective is, precisely, to discover whether or not society is willing to pay an increased price for drugs because they are used for a RD; it is a path to equity through solidarity. In any case, it seems clear that RD patients have the right to be adequately cared for, including treatment (Rombach, Hollak, Linthorst, & Dijkgraaf, 2013). When there is an effective treatment, we have to administer it, invoking when necessary the rule of rescue (referring to the instinctive response to help people whose lives are at risk, regardless of the costs of the action) or legal imperatives. Rejecting an effective orphan drug for economic reasons is not a valid option, according to Luzzatto et al. (2015), who defends the

Many rare diseases are also very serious and highly incapacitating, especially when manifested during childhood.

use of persuasion and creativity to negotiate better economic conditions. But, what is the best way to negotiate with an insurance company or with the administrations that limit or deny orphaned treatments because of their price? (Kesselheim, McGraw, Thompson, O'Keefe, & Gagne, 2015).

«THE ESTIMATED TIME TO RECEIVE A DIAGNOSIS IS FIVE YEARS»

THE TREATMENT FOR DUCHENNE MUSCULAR DYSTROPHY

How do the media represent the complex arguments and counter-arguments used until now in news items concerning access to orphan drugs? Douglas, Wilcox, Burguess and Lynd (2015) lament the times when the media not only reacted to individual calls for help, but when they also explored the debate on the cost, risk and benefits associated with financing these drugs.

To illustrate these types of representations, here we look at the case of an orphan drug for a certain profile of patient with Duchenne muscular dystrophy (in this case, ambulatory patients). It is marketed as *Translarna* and its active substance is ataluren, which contributes to slowing the progression of the disease.

«REJECTING AN EFFECTIVE ORPHAN DRUG FOR ECONOMIC REASONS IS NOT A VALID OPTION»

In Spain, some patients have accessed the treatment through a process known as «compassionate use»: the drug is administered in special cases to these patients, before it is approved, on grounds of urgency and severity.

From January to August 2015, we detected three cases who had problems accessing ataluren who received special media coverage: one in Andalusia, another in the Valencia Region and the third in the Madrid Region. Here we will look more closely at the latter, the case of a boy named Sydney and his family. The drug, developed by *PTC Therapeutics Limited*, obtained commercialisation approval on 31 July 2014.

Ideally, we would also, at minimum, analyse the discourse elements of the case such as time, space, agents, responsibilities and arguments. Nonetheless, for this article we will focus only on the last point, arguments. To this end, we have analysed five news items in different media, as well as an official statement by the Madrid autonomous-region government's Department of Health. The news items, in chronological order, are as follows: 1) «My son would have adequate treatment outside of Madrid» (Bengoa & Sevillano, 2015a). 2) Sydney's mother's statements in *La Sexta Noticias* (Cutillas & Serrano, 2015). 3) «The autonomous region now promises to treat the boy with Duchenne syndrome» (Bengoa & Sevillano, 2015b). 4) «The parents of the boy with Duchenne's now have the medication for their son» (Efe, 2015). 5) «Sydney now has the medication to stop his disease» (Bengoa, 2015). Meanwhile, the statement published on the Department of Health's website read: «The boy with Duchenne's starts his treatment today» (Comunidad de Madrid, 2015).

The key stages in the process, as they appear in the texts, would be: In July 2014 the orphan drug was approved for commercialisation. The family started to ask for it but the hospital where Sydney was receiving treatment denied access to the drug twice, the last time in July 2015. Other children in different Spanish autonomous regions (as well as in other countries) were already receiving the medication months before via compassionate use. The family decided to raise the problem publicly through the media and started collecting support signatures on the digital platform *Change.org*. The Madrid autonomous-region

government's Department of Health intervened and obtained the medication, with the help of the Ministry of Health, in just one day, when the usual procedure takes around one month. On 15 August 2015, just a few days after the case went public in the media, Sydney received the treatment.

We identified the following arguments in the analysed news, depending on whether they favour the request directly or indirectly (proactive), or they oppose it directly or indirectly (reactive). Proactive argumentation is based in the following arguments: firstly, the drug is useful, slows down the disease, is approved by the European Medicines Agency and was requested by Sydney's doctor; secondly, the medication is already in use in other children in other autonomous regions thanks to compassionate use; and thirdly, although the treatment is expensive, only a reduced group of 10 % of the 400 cases in Spain can receive the treatment, so the expenditure will be exceptional and limited.

On the other hand, reactive argumentation and its demonstrative elements also appear in media discourse. The arguments that support it are: firstly, the medicine does not cure the disease; secondly, it cannot be bought in Spain yet (although it is approved by the Spanish Agency of Medicines, the hospital does not respond to these types of requests); and thirdly, it is too expensive: more than 300,000 euros per year. Furthermore, all of this transpired in an era of compulsory budget cuts.

Therefore, in a single case, we can find the basic arguments and counter-arguments relating to the access to expensive orphan drugs. But emotion often overcomes rationality, which should take precedence in journalistic information. Let us pause for a moment on the first reactive argument: the drug does not cure the disease. They said about Sydney's medicine that «it does not cure [the disease], but it does slow down [its] progression» (Bengoa & Sevillano, 2015a) or that «it delays the progression of this type of muscular dystrophy» (Bengoa & Sevillano, 2015b). As we have stated before, improving quality of life, slowing down or stopping the progression of a disease is not a minor effect when we discuss RDs.

La madre de Sydney, a la salida del hospital 12 de Octubre con el medicamento para su hijo. / KIKE PARA

Sydney ya tiene la medicina para frenar su enfermedad

AITOR BENGOA. **Leganés**

Los padres de Sydney, el niño de Leganés que padece distrofia muscular de Duchenne, acudieron ayer al hospital 12 de Octubre para recoger el fármaco especial Ataluren, que puede frenar los síntomas de esta enfermedad rara y actualmente incurable.

La Comunidad de Madrid se lo había negado en dos ocasiones, a pesar de que otras comunidades españolas ya lo dispensaban. Gracias a la movilización de la familia, el pequeño empezará hoy su tratamiento. Su madre, emocionada, ha atribuido su logro a la fuerza del "amor y la voluntad".

Sydney es un niño de cinco años que vive en Leganés al que le chiflan los regalices rojos y la música. Su agudo sentido del oído detecta cadencias y ritmos ocultos en los sonidos que le rodean, que él sigue con precisos tarareos y divertidos bailes que interrumpe solo para sonreír. Una de sus canciones favoritas es *Back to Black*, de Amy Winehouse. También disfruta convirtiendo sus dedos en pinceles, la salsa de los macarrones que cocina su abuela en témpera y la pared de la cocina en un improvisado lienzo en el que plasma su abstracto arte.

El gusto por pintar es una cualidad que comparte con su madre, Ana Isabel López, de 39 años, quien pocas horas antes se emocionaba al salir del hospital 12 de Octubre junto a su marido, Cristóbal Escudero, aferrada a un bolso en el que lucía la cara de Sydney. Dentro estaba el fruto de 1,5 años de lucha: el Ataluren, un fármaco que puede ayudar a su hijo a frenar los síntomas de la distrofia muscular de Duchenne, dolencia que padece desde que era un bebé.

Se trata de una enfermedad rara que no tiene cura. Los afectados pierden paulatinamente sus funciones musculares. En torno a los 12 años han de utilizar silla de ruedas y su esperanza de vida es de unos 30. Solo afecta a varones. En España hay unos 400 pacientes. El Ataluren, que no está comercializado y que solo se puede conseguir con permiso especial en un hospital, está indicado para un pequeño grupo que presenta una mutación genética particular, que es el caso de Sydney.

"Hoy es uno de los días más felices de nuestra vida", explicaban ayer en su casa los padres porque, aunque son conscientes de que la enfermedad es incurable, el nuevo fármaco mejorará la calidad de vida de su hijo. Obtenerlo no ha sido fácil para ellos, que actualmente, además, están desempleados e hipotecados. Hasta en dos ocasiones la Comunidad de Madrid se lo negó, a pesar de que en otras comunidades autónomas se administraba esta medicina, que puede llegar a costar unos 300.000 euros.

Gracias al consejero

Todo cambió esta semana, cuando el caso saltó a los medios de comunicación tras la movilización de la familia del pequeño, que ha contado con el apoyo del Ayuntamiento de Leganés, asociaciones y miles de personas que han respaldado su petición con firmas en la web change.org. "Gracias", ha repetido una y otra vez la madre, nerviosa, a la salida del hospital. Ha extendido el agradecimiento al consejero de Sanidad madrileño, Jesús Sánchez Martos, que se implicó personalmente para agilizar el proceso de adquisición del Ataluren, que ha tardado 24 horas. López ha pedido, asimismo, que se investiguen esta y otras enfermedades raras.

Hoy, al despertar, su hijo empezará a tomar por fin los sobres que le han prescrito. Tres al día, diluidos en agua o yogur. Hasta entonces, las estrellas y el koala que su madre —apasionada de Australia— ha pintado en las paredes de su habitación velarán, como cada noche, por los sueños de Sydney.

Above and on the right, one of the news items analysed in the article. The woman is Sydney's mother, a five-year-old boy with Duchenne muscular dystrophy. She is carrying the medication to slow down her son's disease, which they had been denied twice before.

«IMPROVING QUALITY OF LIFE, SLOWING DOWN OR STOPPING THE PROGRESSION OF A DISEASE IS NOT A MINOR EFFECT WHEN WE DISCUSS RARE DISEASES»

A POLARISED DEBATE OF BLACKS AND WHITES

Our analysis concludes that the RD debate is currently polarised. It focuses, on the one hand, on the price of drugs and on the other, on their curative efficacy, without taking into account other positive effects of these medications beyond strictly curing. In times of crisis, with such an important issue as

ELPAÍS **3**

MADRID

La Comunidad promete ahora tratar al niño con síndrome de Duchenne

A. B. / E. G. S., **Madrid**
La Comunidad de Madrid anunció ayer que "facilitará" a Sydney, el niño de cinco años de Leganés que padece una enfermedad rara llamada síndrome de Duchenne, el fármaco experimental Ataluren, que retrasa la progresión de este tipo de distrofia muscular. Esta promesa se produce horas después de que los medios de comunicación publicaran que el hospital 12 de Octubre, dependiente de la Consejería de Sanidad, le estaba negando la medicación. Niños con la misma dolencia que Sydney están recibiendo este tratamiento en otras comunidades autónomas, motivo —la equidad de acceso a la salud— que su madre alegaba para que se lo facilitaran también en la región.

La Comunidad de Madrid emitió una nota de prensa en la que niega que el fármaco se le hubiera denegado. Documentación consultada por EL PAÍS muestra que no es así. El médico del niño solicitó el Ataluren como uso compasivo en marzo de 2015. La Comisión de Farmacia del hospital no lo autorizó, alegando que el fármaco aún no se comercializa en España. La Agencia Europea del Medicamento lo ha aprobado y ya se comercializa en otros países europeos. "Es criterio general de la dirección del hospital no autorizar la adquisición de medicamentos que se encuentran en esa situación", asegura la respuesta.

Criterio: "No autorizar"
A Ana Isabel López, la madre de Sydney, le denegaron una segunda vez el tratamiento. En un documento fechado el 15 de julio, la directora gerente del hospital reconoce que "es posible solicitar" la importación del fármaco como medicamento extranjero —así se ha hecho en otras comunidades—, pero que el criterio del hospital es el de "no autorizar" la compra de este tipo de fármacos. El documento alude para denegarlo al Real Decreto 16/2012, en el que el Gobierno central incluyó recortes en la asistencia sanitaria, entre ellos el copago de los medicamentos de los pensionistas y la exclusión de los inmigrantes sin papeles.

El comunicado de la Comunidad menciona asimismo que al niño se le administrará el fármaco "de acuerdo con criterios clínicos y atendiendo a la solicitud de la doctora" que le atiende. El consejero, Jesús Sánchez Martos, dijo a EL PAÍS que ha citado a los padres del niño a su despacho este miércoles y que ha dado orden al hospital para autorizar el fármaco. "No se tendría que haber denegado", añadió.

the life of patients, the debate tends to be simplified into blacks and whites, avoiding the shades of grey. Questions such as these are forgotten: Is society aware of the investment necessary to implement any medication? Do we have information on clinical trials and the research process in general? Are we trained to use our health system so investments reach the most important issues? Do we know what RDs are and what their specific medical and psychosocial characteristics are? How do the media contribute to knowledge of these issues?

We should keep all of these questions more present when we discuss health-care models. In any case, they can only be answered if we set up an adequate health training system for the citizenry with the help of the media.

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