LEXICAL-TERMINOLOGICAL EVOLUTION OF RARE DISEASES: REVIEW OF DICTIONARIES OF THE REAL ACADEMIA ESPAÑOLA

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■ ABSTRACT: This paper focuses on the terminological development of fifty-four rare diseases. It analyzes the degree of lexical acceptance that these terms have and have had –over time– in their treatment and inclusion in academic dictionaries of Spanish language. Until now, only twenty of the selected terms have been included in DRAE, accounting for 37.03 %. This situation shows the lack of solid criteria in incorporating new terms about rare diseases in this general dictionary. The most noticeable case is fibromyalgia, a pathology that has recently ceased to be considered a rare disease, which, however, does not appear in DRAE. If, on the one hand, it must be assumed that the DRAE is not a specialized medical dictionary; on the other, it must assess the degree of incorporation of these voices in social life.

■ KEYWORDS: Vocabulary. Terminology. Diccionario de la Real Academia Española. Rare diseases.

Introduction: rare diseases

The study of less prevalent pathologies, in its various approaches, is gaining prominence in recent years; this is mainly due to the activities of the Federación Española de Enfermedades Raras (FEDER). Rare pathologies are considered to be those whose prevalence rate is less than five per ten thousand inhabitants (EUROPEAN COMMISSION, 2008). It is further estimated that there may be somewhere between six and eight thousand diseases of this kind, many of them of genetic origin and chronic nature. The percentage of population affected by these ailments is estimated at about 7 %. Therefore, there would be three million people with this problem in Spain (ENFERMEDADES RARAS, 2009).

Furthermore, being so rare, they have the disadvantage of an unknown etiology resulting in disorientation in the diagnostic process, late or absent in most cases.
Consequently, not enough information on the care and treatment that can improve the quality of life of these people (POSADA DE LA PAZ; GROFT, 2010) is provided. Paradoxically, as a whole, those affected by these ailments constitute a large group, but have been practically invisible until just a decade ago (MATEO, 2001; GONZÁLEZ-MENESES et al., 2008). And apart from the lack of legal cover to promote research, there is also a marked deficit of drugs specifically designed to alleviate such diseases (ENSERIO, 2009).

Similarly, they present the perverse picture of low commercial return, which directly relates to the shortage of means for treatment, although this may be disputed with the argument that research on rare diseases positively impacts the understanding of common ailments (GRIGGS et al., 2009). It is evident that those rare diseases that have attracted public attention for some time are currently benefiting from research and health care. This progress has resulted in a quantitative and qualitative improvement of life of the patients, who achieved greater public awareness of this issue.

It so happens that the number of scientific publications on rare diseases has increased in recent years, especially from a medical approach. The communication sphere has likewise been interested in investigating the health context, since the end of last century. Thus, a current named health communication was born in the Anglo-Saxon area. An example we cite is the recognized Gwyn’s manual (2001). Meanwhile, the Hispanic world echoes this relatively nascent line of research, as evidenced by the birth of the scientific journal Comunicación y Salud in 2011, as well as the publication of the book Lenguaje, comunicación & salud (FORNIELES; URÁN, 2011).

When focusing on the study of less prevalent diseases from a linguistic angle, sources to consult encompass various works on the communicative profile (BAÑÓN et al., 2011; FORNIELES; URÁN, 2011), the discursive presentation of these conditions in the press (RIDAO, 2011; ARCOS, 2013), as well as the analysis of pathological discourse in specific diseases, such as Asperger syndrome (RODRÍGUEZ MUÑOZ, 2013) or Moebius syndrome (REQUENA, 2011).

**Lexical-terminological issues: specialized idioms**

Due to the nature of this research, which combines both the lexicological and terminological orientation, the scope of analysis should be comprehensive, so that it reflects critically on the lexicographical treatment that the names of fifty-four diseases that have been studied received and whether they have been included in general or only dictionaries specialized in the field of medicine, a question inherent to the debate on the difficult coexistence of the terminology of technolects in general dictionaries.

Regarding the dilemma on the existence of specialized languages, this paper supports the thesis that they do exist, though completely interwoven in the social life of the speakers. Hoffmann (1998) considers that specialized languages relate to
the linguistic resources characteristic of a particular communicative domain marked by a common specialty, and one that seeks to facilitate communication between professionals of a specific area. Mejri (2012, p.22) defines specialized discourse in a strictly linguistic approach: “[…] il s’agit d’un discours qui se caractérise par une hétérogénéité structurelle où coexistent un discours appartenant à la communication courante et des segments spécialisés inintelligibles pour les non experts”\textsuperscript{1}.

It would be wrong to reduce a specialized language purely to terminology, since it uses both terms, specialist designations, as well as nonlinguistic symbols in expressions where regular resources of a particular language are used; therefore, “[la lengua especializada] se puede definir como el uso de una lengua natural para exponer técnicamente los conocimientos especializados”\textsuperscript{2} (LERAT, 1997, p.18).

Apart from that, the list of pathologies selected a priori vies for specialised language, even though many of them are part of the general language as a result of great frequency of usage. Rondeau (1981) and, more recently, Cabré (2010) warn of the difficulty of drawing a sharp distinction between common language and specialized vocabulary. Moreover, Cabré (1999), while referring generally to any field relating to knowledge or language, notes that it is almost impossible to discriminate between what is to be considered general and what is specific. The fact that these terms pertain to a specialized area on the one hand, but on the other, are also used in common parlance, results in such distinction being rather blurred (SANTAMARÍA, 1998).

In the introduction to the work by Mogorrón and Mejri (2012, p.13), a “consubstantial continuity” between general language and specialized languages is postulated. Furthermore it is argued that the specialized discourse follows the rules of the general language and settled that specialization should be conceived as a scale allowing for situating popularized texts in an intermediate position between the specialized and general.

This difficulty of placing the terms that belong to both the specialized language and the common language has implications for general dictionaries, as is the case of academic ones, so that inaccuracies are reflected in the incorporation of these lexical units that can be doubly regarded as specialized lexicon or common vocabulary (CABRÉ et al., 2001), as with the medical and health sciences.

The issue of incorporation of specialized vocabulary in general dictionaries has been argued over extensively. Although, as pointed Anglada (1992), no solid criteria exists for a lexicographer to shed light on, firstly, the quantitative aspects of the terms to be featured, and, secondly, the qualitative aspects concerning how to collect and define such lemmas.

\textsuperscript{1} “[…] A discourse characterized by structural heterogeneity where the currently common communicative discourse coexists with specialized segments unintelligible to the uninitiated”. Mejri (2012, p.22).

\textsuperscript{2} “(The specialized language) can be defined as the use of a natural language to disclose technically a specialised knowledge”. (LERAT, 1997, p.18).
As Gutiérrez Rodilla affirms (2005), the social life of individuals is influenced by the repercussions and applications of science, so that science and technology constitute a primary cultural element:

It follows that society needs good materials for promoting science, including specialised informational dictionaries that, besides enabling general dictionaries and lexicons to be free from much of the scientific terminology, present information related with this vocabulary in a manner suitable for people who are not specialists in each of the subjects. That is to say, dictionaries able to find the balance between scientific pressure and general accessibility.³ (GUTIÉRREZ RODILLA, 2005, p.30).

In the Hispanic world, the first 1983 edition of the Vocabulario científico y técnico published by the Real Academia de Ciencias Exactas, Físicas y Naturales operates with a selection of registered lexicon, consequently it should be classified among specialized dictionaries, versus general or global ones (CUESTA MARTÍNEZ; DE VEGA PÉREZ, 1992). The second edition, dated 1990, describes itself as a vocabulary of mixed character straddling terminological and encyclopedic information.

On the other hand, Lerat (1997) reports that the Trésor de la langue française, while describing medical terms, employs four procedures characteristic of general dictionaries: (1) An indicator on the area of expertise. The use of a label MEDÉCINE (medicine) constituting a technical connotation of a technical dialect. (2) The use of analogy. (3) A definition for patients. (4) Absence of entry for the syntagmatic compound. This section notes that the multiple volume dictionaries do not collect more entries than those contained in a single volume, but that the quantitative difference is due to the amount of information included in each entry, not the existence of a greater number of lemmas.

Additionally, making the mistake of encyclopedism in lexicographic definitions is to be avoided. Hence Bosque (1982) warns of the often encountered impossibility of foregoing encyclopedic definition in dictionaries, mainly because it benefits readers. This occurs with median frequency when defining nouns. Accordingly, the difficult task of marking the boundaries between lexicographical and encyclopedic definitions is emerging as one of the great and interesting dilemmas inherent in modern lexicography.

³ “De ahí se desprende que la sociedad necesite buenos materiales de divulgación científica, entre los que deben encontrarse los diccionarios especializados divulgativos que, además de permitir a los diccionarios generales y a los de lengua liberarse de una buena parte de la terminología científica, presenten la información relacionada con ese vocabulario de una manera adecuada para las personas que no son especialistas en cada una de las materias. Es decir, diccionarios que sepan encontrar el equilibrio entre la presión científica y la accesibilidad general”. (GUTIÉRREZ RODILLA, 2005, p.30).
Methodology

This modest study aims to investigate the terminological profile from a lexicographical point of view, as seen from the presence of lexical units relating to certain rare diseases or ones of low prevalence in successive editions of the dictionary of the *Real Academia Española* (RAE, 2001). The time frame is established from the first installment of the glossary, *Diccionario de autoridades* (RAE, 1726-1739), to the latest modifications to the entries of the twenty-third edition underway in the online consultation directory⁴. Thus, not only the presence or absence of entries devoted to less prevalent ailments in academic dictionaries will be addressed, but we will also try to trace a brief outline of possible variations in defining them.


This list was checked and supplemented from the base of low-prevalence diseases from the *Portal de información de enfermedades raras y medicamentos huérfanos* (Orphanet), also with online access through website⁷. Apart from the thirty-three rare diseases listed above, the following twenty-one will be considered: “anodontia”, “anotia”, “argyria”, “botulism”, “brucellosis”, “blepharospasm”, “brachydactyly”, “cryptophtalmia”, “dengue”, “diphtheria”, “exencephaly”, “favism”, “fibrosarcoma”, “gigantomastia”, “glioma”, “hemophilia”, “hypertrichosis”, “ichthyosis”, “legionellosis”, “leprosy” and “microcephaly “.

In the selection of these fifty-four words, we decided to forego the complex lexical units that cannot do without the phrases “disease / syndrome / disorder”, i.e. those that have no integrity to form by themselves simple and autonomous lexical units (eg, “Fabry disease” or “chronic fatigue syndrome”). Those units which are necessarily specified by some other modifier (as in the case of “juvenile chronic arthritis”) and in general, all those that contain proper nouns (like “Fanconi anemia”) are not included either.

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⁴ RAE (2001).
⁵ FEDER (2014).
⁶ It is to be noted that fibromyalgia has recently stopped being considered a rare disease, though it is catalogued as such in the description.
⁷ ORPHANET (2014).
For technical definitions of each of the terms referring to rare diseases we have consulted mainly the Orphanet website\(^8\), accessible through the Registro Nacional de Enfermedades Raras del Instituto de Salud Carlos III (2014), as well as the specialized dictionary Dicciomed\(^9\).

Analysis and results

The first classification is set according to whether or not these words appear in various editions of DRAE to then we further investigate the information provided on each specific condition.

Terms with definition

This section provides a critical analysis of the twenty rare diseases, in alphabetical order, that are collected in the different editions of DRAE.

_Achondroplasia_

The disease is defined for the first time in the nineteenth edition of DRAE (RAE, 1970), accompanied by the designation _Pat._ (Pathology). The only variation of the definition in the twenty-second edition is the substitution with the designation of the scientific-technical discipline: _Med._ (Medicine).

It is defined as a “[…] variedad de enanismo caracterizada por la cortedad de las piernas y los brazos, con tamaño normal del tronco y de la cabeza y desarrollo mental y sexual normales […]”\(^10\) (RAE, 1970, p.18). Indeed, one feature of this type of dwarfism are short limbs; however, the clinical feature of macrocephaly, or moderately disproportionate size of the head relative to the body, is not specified.

_Acromegaly_

The first definition for this condition appears in the sixteenth edition of DRAE (RAE, 1936), is repeated at the seventeenth (RAE, 1947), in the eighteenth (RAE, 1956) and, from the nineteenth edition (RAE, 1970), the designation _Pat._ _is_ included and kept in the twentieth (RAE, 1984) and in the twenty-first (RAE, 1992) edition. In the last issue, along with the designation _Med._, some changes concerning primarily etiology occur.

\(^8\) ORPHANET (2014).
\(^9\) DICCIOMED (2014).
\(^10\) “Variety of dwarfism characterized by the shortness of the legs and arms, with normally sized head and torso, as well as a normal mental and sexual development”. (RAE, 1970, p.18).
Generally, it is treated as a “enfermedad crónica […] que se caracteriza principalmente por un desarrollo extraordinario de las extremidades”\textsuperscript{11} (RAE, 1936, p.20). By contrast, it is attributed to “la lesión de la glándula pituitaria”\textsuperscript{12} in the pre-2001 editions, while in the latter ones, it is explained by “un exceso de secreción de hormona de crecimiento por la hipófisis”\textsuperscript{13} (RAE, 2001). Although this supposes an advancement of the definition, a more explicit mention of gigantism is still missing (as there is reference to dwarfism in achondroplasia), although it does come announced by hypertrophy of the extremities in any case.

**Albinism**

This disease is defined in the twelfth edition of Academy’s dictionary (1884). The entry remained unchanged to the twenty-second edition (RAE, 2001), “quality of albino”. One wonders, therefore, why this one continually redirects to the adjective albino when the definitions of other diseases do not. One of the most plausible causes may be that this word is first defined in the Diccionario de autoridades (RAE, 1726, p.169) as “[…] la persona que nace con los cabellos y carne de extremada blancura, por cuya causa ve muy poco, y menos cuanto más luce el Sol y cuanto está más cerca de la luz.”\textsuperscript{14} This is obviously a prescientific definition already modified in the second revised and expanded edition of this dictionary where it appears as “[…] el que de padres negros, o de casta de ellos, nace muy blanco y rubio, conservando en lo corto y retortijado del pelo y en las facciones del rostro las señales que tienen los negros y los distinguen […].”\textsuperscript{15} (RAE, 1770, p.139). It is difficult to assess to what extent this definition is a step forward from the prescientific character of the earlier, as it appears to be completely unscientific.

What is certain is that this definition is incorporated in the first edition of DRAE and that it does not change until the twelfth edition, when the scientific description of this condition emerges: “[…] falto entera o parcialmente, y por anomalía congénita, del pigmento que da a ciertas partes del organismo los colores propios de cada especie, variedad o raza y, por tanto, con la piel, el iris, el pelo, el plumaje, etc. más o menos blanco. Dícese del hombre y del animal […]”\textsuperscript{16} (RAE, 1884, p.43). This is, basically, the definition that is preserved almost intact to the twenty-second edition of DRAE (2001), where it appears somewhat remodeled as “Dicho de un ser vivo: Que presenta ausencia

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\textsuperscript{11} “Chronic disease […] which is mainly characterized by an extraordinary development of limbs”. (RAE, 1936, p.20).
\textsuperscript{12} “The injury of the pituitary gland”.
\textsuperscript{13} “Excessive secretion of growth hormone by the pituitary gland”. (RAE, 2001).
\textsuperscript{14} “A person who is born with hair and flesh of extreme whiteness and for that sees very poorly, and even less when the sun shines and the closer to the light it gets”. (RAE, 1726, p.169).
\textsuperscript{15} “One from black parents, or their kind, very white and ruddy, but that conserves signs by which blacks are distinguished: short curly hair and facial features”. (RAE, 1770, p.139).
\textsuperscript{16} “Lacking, in whole or in part and as a congenital anomaly, in pigment that gives certain parts of the body the coloring characteristic of each species, variety or race, and thus with skin, iris, hair, plumage, etc. more or less white. It is used for man and animal”. (RAE, 1884, p.43).
congénita de pigmentación, por lo que su piel, pelo, iris, plumas, flores, etc., son más o menos blancos a diferencia de los colores propios de su especie, variedad o raza.”17

It might be that a redesign of this entry, where there is no explicit reference to melanin deficiency and no mention of the genetic, not only congenital, character of this disease, is in order.

**Botulism**

First appears in the 1970 DRAE referring to a disease caused by a toxin of a certain bacillus contained in food that is not properly conserved. There is no apparent change to this definition in the twenty-second edition.

**Brachycephaly**

Here again is a definition redirected to another term, brachycephalic, defined as “la persona cuyo cráneo es casi redondo porque su diámetro mayor excede en menos de un cuarto al menor”18, appeared in the thirteenth edition of DRAE (RAE, 1899, p.1046). It has been preserved without change to the twenty-second edition of the Academy’s dictionary.

In effect, some scientific character is entailed, but it is a somewhat confusing explanation of the symptoms of a cranial deformation, basically characterized by a short, broad head with a flattened occipital part (back of a head).

**Brucellosis**

In 1989 the Dictionary of the Real Academia Española includes among its entries the brucellosis in order to define an infectious disease transmitted to humans by various animals and caused by microorganisms; Malta fever, Mediterranean fever, etc. In the twenty-second edition, the reader is offered a slightly more explicit definition which differentiates between the types of animals that can transmit the infection as well as the type of bacteria. The definition is: “Enfermedad infecciosa producida por bacterias del género Brucella y transmitida al hombre por los animales domésticos […]”19 (RAE, 1989, p.238).

**Dengue**

In the edition of 1732 there are two entries for this word. The first defined dengue as “melindre mugeril que consiste en afectar damerías, esguinces, delicadezas, males y a veces disgusto de lo que mas se suele guitar. Es voz modernamente inventada

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17 “Said of a living being: that has congenital absence of pigmentation, so that its skin, hair, iris, feathers, flowers, etc., are more or less white as opposed to the usual coloring of its species, variety or race”. (RAE, 2001).
18 “The person whose skull is nearly round because its larger diameter exceeds the lesser by less than a quarter”. (RAE, 1899, p.1046).
The second entry refers to dengue as a female garment. In 1899, in the supplement to the *DRAE* specifically, the definition for dengue appeared for the first time in medical terms as “[…] *enfermedad febril, epidémica y contagiosa, que se manifiesta por dolores de los miembros y un exantema semejante al de la escarlatina* […]” (RAE, 1899, p.321). Already in 1914 the two definitions with which the word was introduced appear, in addition to the medical definition included in the supplement of 1899. In 1925 there is a new entry for dengue with two definitions specific to Chile. The first refers to a plant and the second, to this plant’s flower.

The 1983 edition also includes an illustration. And in 1992 a new definition reads concerning the first entry for the word. The entries from 1732, commented ever since, take first and second place. A new one appears in the third: “swagger”; and fourthly the medical definition, not classified with *Med.* any longer, but with *Pat.* In the twenty-second edition the classification *Med.* is restored, as well as the third place, leaving the fourth place to “swagger”.

**Diphtheria**

Its first appearance dates to 1884 when it was defined as “[…] *enfermedad caracterizada por la formación de falsas membranas, que se observa en las mucosas y en la piel desprovista de epidermis* […]” (RAE, 1884, p.385). In 1899, a more precise localization of the disease is offered within this definition, since the one offered in 1884 is complemented by “common in the throat”. This definition will remain for twenty-eight years, until a less explicit description is offered in 1927 but which adds the feature of “infectious disease”. In 1936 there is a more comprehensive definition that describes diphtheria as infectious and contagious disease and discloses some symptoms of the disease: “fever and prostration”. The 1950 edition presents a brief definition of diphtheria which includes only its infectious nature and the formation of false membranes on the mucosae. Six years later, in 1956, the definition from 1936 is recovered and stands for the next twenty-seven years, for in 1983 the entry from 1950 reappears. A year later, in 1984, the definition of 1936 is taken up; and in 1989 one from 1936, which is to remain in force until the twenty-second edition.

**Scleroderma**

This term is defined as “*enfermedad crónica de la piel caracterizada por el abultamiento y la dureza primero, y por la retracción después*” in 1927, in the

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20 “Female fussiness consisting of affecting fragility, delicacy, hurt and sometimes disgust for what usually bothers one most. It is a modernly invented word”. (RAE, 1726-1739, p.69).
21 “Febrile illness, epidemic, contagious, manifested by aching members and a rash similar to the one from scarlet fever”. (RAE, 1899, p.321).
22 “Disease characterized by the formation of false membranes, which is observed in the mucosae and skin devoid of epidermis”. (RAE, 1884, p.385).
23 “Chronic skin disease characterized by the swelling and hardness first, and then retraction”. (RAE, 1936, p.863).
Diccionario manual e ilustrado de la lengua española by Real Academia Española (RAE, 1927, p.863). Later, it will be included in these terms in the sixteenth edition of DRAE (1936) and remains to this day.

It seems advisable to specify the etiology of this disease as an accumulation of collagen in the skin and other organs, as well as extending the symptomatic spectrum beyond just skin issues, to mention also the muscular and skeletal manifestations, or even further to internal organs.

**Phenylketonuria**

Without referring to previous editions, the twenty-second edition of the academic dictionary takes a step forward in defining the disease as a “[…] anomalía hereditaria que consiste en la alteración del metabolismo de la fenilalanina, que puede provocar retraso en el desarrollo y deficiencia mental […]”24 (RAE, 2001).

**Glioma**

The word glioma is defined for the first time in the twenty-second edition of DRAE referring to a glial cell tumor.

**Hemophilia**

It was first included in the DRAE (1936, p.677) as “[…] hemopatía hereditaria, caracterizada por la dificultad de coagulación de la sangre, lo que motiva que las hemorragias provocadas o espontáneas sean copiosas y hasta incoercibles […]”25, and not until the current edition (the twenty-second) has there been modification, minimal at that, with respect to the vocabulary used: the word “incoercible” is replaced by “difficult to quench”.

**Hydrocephalus**

The term hydrocephalus is presented firstly in the fifteenth edition of DRAE (1925) as “dropsy of the head” and this definition remains unchanged until the twenty-second edition (RAE, 2001) where it is announced as “dilatación anormal de los ventrículos del encéfalo por acumulación de líquido cefalorraquídeo”26. This is obviously a definition that incorporates, in this case, sufficient scientific nuance and, from a lexicographical point of view, minimizes the effect of circularity by omitting the term dropsy, equivalent to an accumulation of serous fluid above typical levels.

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24 “Hereditary anomaly that alters the metabolism of phenylalanine, which can cause stunted development and mental retardation”. (RAE, 2001).

25 “An inherited blood disorder characterized by difficulty in blood clotting, which occasions a spontaneous or provoked bleeding to be copious, even”. (RAE, 1936, p.677).

26 “Abnormal dilation of the brain ventricles by accumulation of cerebrospinal fluid”. (RAE, 2001).
**Ichthyosis**

The definition of ichthyosis offered by RAE in the 1927 edition, one that will remain for the next several editions, refers to it as “enfermedad de la piel, que se caracteriza por cubrirse ésta de un tejido escamoso”\(^{27}\) (RAE, 1927, p.1079). However, in the twenty-second edition, ichthyosis is defined as “alteración patológica de la piel, que toma aspecto escamoso”\(^{28}\) (RAE, 2001).

**Legionellosis**

It is first recorded in the DRAE in 2001, referring to a “enfermedad causada por bacterias del género Legionella, que se difunde especialmente por el agua y por el uso de nebulizadores”\(^{29}\).

**Leprosy**

It is included in the 1734 edition as a “[…] especie de sarna que cubre el pellejo del cuerpo humano con unas costras mui feas, por partes blancas y por partes negras. Hace mui áspero el cutis, y va comiendo las carnes con vehemente comezón. Es voz puramente latina […]”\(^{30}\) (RAE, 1726-1739, p.386). The definition does not change in 1780.

In 1803 leprosy is defined as a “[…] enfermedad cutánea y contagiosa que consiste en unas pústulas hediondas, arracimadas y escamosas, que se van extendiendo por todo el cuerpo, y termina en una fiebre lenta […]”\(^{31}\) (RAE, 1803, p.512). This one will last until the 1899 edition when some common symptoms are offered within the definition of leprosy: “[…] enfermedad transmisible por herencia y de larga duración, que se manifiesta por manchas generalmente de color leonado, tubérculos insensibilidad de la piel, ulceraciones y caquexia […]”\(^{32}\) (RAE, 1899, p.597).

In 1925 the difficulty in curing this disease is alluded to. In the edition published in 1936 there is a new definition for leprosy in addition to the one which had previously stood since 1899, with some amplification in 1925. This new definition refers to leprosy as a “[…] enfermedad, principalmente de los cerdos, producida por el cisticerco de la tenia común, y que aparece en los músculos de aquellos animales en forma de

\(^{27}\) “Skin disease, characterized by a scaly covering”. (RAE, 1927, p.1079).

\(^{28}\) “Pathological alteration of the skin, taking a scaly aspect”. (RAE, 2001).

\(^{29}\) “Disease caused by a bacteria of the Legionella genus, which spreads through water and the use of vaporizers”. (RAE, 2001).

\(^{30}\) Kind of scabies that covers the skin of the human body with very ugly scabs, partly white and partly black. It makes the skin very coarse and consumes the flesh with vehement itching. It is a purely Latin word. (RAE, 1726-1739, p.386).

\(^{31}\) “Contagious dermal disease that consists of stinking bulging scaly pustules that spread all over the body, and ends in a slow fever”. (RAE, 1803, p.512).

\(^{32}\) “A long lasting disease transmitted by inheritance, manifested by usually tawny marks, tubers, insensitivity of the skin, ulcerations and cachexia”. (RAE, 1899, p.597).
pequeños puntos blancos […]” (RAE, 1936, p.770). This edition includes the onset of the disease and its form in animals.

The term cachexia contained in the definition of leprosy since 1899 will not be replaced by anesthesia until the edition published in 1956 and the adjective tawny characterizing the color of the marks disappears, leaving the leprosy defined as follows: “[…] enfermedad infecciosa crónica, caracterizada principalmente por síntomas cutáneos y nerviosos, sobre todo tubérculos, manchas, úlceras y anestesias […]” (RAE, 1956, p.798).

From 1734 onwards, this word is not listed in any field until 1970, when both definitions are classified as pathology Pat. In the 1984 edition, particularly the manual volume IV (incognito-paper), anesthesia is substituted for the “falta de sensibilidad en la zona afectada”35. As for the second definition, it gets classified with Veter., since it is specific to animals. This categorization will be retained in the next edition of 1989. However, in 1992 the word anesthesia is restored in the definition, as well as the use of Pat. for the categorization of the second definition. In the last edition, the only change from the previous is the category of the second definition. Veter. is again used instead of Pat.

Microcephaly

In the entry from 1936 it is described as “quality of microcephalic”. The word microcephalic is defined in these terms: “[…] dicese del animal que tiene la cabeza de tamaño menor del normal en la especie a que pertenece; y en general, que tiene la cabeza desproporcionada por lo pequeña, con relación al cuerpo […]”36 (RAE, 1936, p.845).

It is in 1950 when the word “microcephalic”, referred to in the dictionary entry “microcephaly”, offers a not so very explicit definition: “of a small head”. Six years later, in 1956, the definition provided of microcephalic in the edition of 1936 is returned to. In 1989 the definition proposed by the Real Academia Española takes up the description “of a small head” again.

In the twenty-second edition of the dictionary (RAE, 2001) there are two definitions for microcephalic, which summarize the definitions given in previous editions. The first one says: “Dicho de un animal: Que tiene la cabeza de tamaño menor del normal en la especie a que pertenece”37. And the second: “Que tiene la cabeza desproporcionada, por lo pequeña, con relación al cuerpo”38.

33 “Disease, primarily of pigs, caused by cysticerci of common tapeworm that appears in the muscles of these animals as small white spots”. (RAE, 1936, p.770).
34 “Chronic infectious disease characterized primarily by cutaneous and nervous symptoms, especially tubers, spots, ulcers and anesthesia”. (RAE, 1956, p.798).
35 “Lack of sensation in the affected area”.
36 “Said of an animal that has a head smaller than normal for the species to which it belongs; and generally, having a disproportionately small head, relative to the body”. (RAE, 1936, p.845).
37 “In an animal: that has a head smaller than normal for the species to which it belongs”. (RAE, 2001).
38 “That has a disproportionately small head, relative to the body”. (RAE, 2001).
Narcolepsy
This condition, also known as “Gelineau syndrome” or “sleep epilepsy”, was not included until the twenty-second edition of the dictionary where it is defined as a “estado patológico caracterizado por accesos irresistibles de sueño profundo”39 (RAE, 2001). Rather than a pathological condition, it ought to refer to a neurological disorder, even though its most characteristic symptoms are, in fact, episodes of uncontrolled and sudden drowsiness.

Nevus
This term is also incorporated for the first time in the twenty-second edition of the Academy’s dictionary, being defined as “alteración congénita muy localizada de la pigmentación de la piel, generalmente de color marrón o azulado”40 (RAE, 2001). It is a proliferation of melanin cells in the skin and mucous membranes that causes localized marks.

Thalassemia
In the twentieth edition of DRAE (1984, p.1281), it is defined as “[…] cualquiera de las anemias hemolíticas hereditarias, que se presentan de modo preferente en individuos de países mediterráneos y se deben a un trastorno cuantitativo en la producción de hemoglobina”41. This definition is maintained until 1992. It will be the twenty-second edition in which there is change in form, not content: “Anemia hemolítica hereditaria, que se presenta de modo preferente en individuos de países mediterráneos y se debe a un trastorno cuantitativo en la producción de hemoglobin […]”42 (RAE, 2001). In any case, it is a complete definition, from a medical perspective for it combines data on the type of the disorder, its etiology, even its geographically localized incidence (in fact, this disease is also known as “Mediterranean anemia”).

Terms not defined
Hereafter the pathologies lacking entry in the various editions of DRAE are included, adding up to a total of thirty-four diseases of low prevalence.

Aniridia
Interestingly, aniridia, an ailment that can have such a simple definition as “clinical absence of the iris”, does not gain an entry in the Academy’s dictionary.

39 “Pathological state characterized by irresistible bouts of sleep”. (RAE, 2001).
40 “Highly localized congenital disorder of pigmentation of the skin, usually of a brown or blue color”. (RAE, 2001).
41 “Any hereditary hemolytic anemia that occurs primarily in individuals from Mediterranean countries and is due to a quantitative disorder in hemoglobin production”. (RAE, 1984, p.1281).
42 “Hereditary hemolytic anemia, which occurs primarily in individuals from Mediterranean countries and is due to a quantitative disorder in hemoglobin production”. (RAE, 2001).
Anodontia
This term has no place in DRAE even if it constitutes a “enfermedad congénita rara que se caracteriza por la ausencia de un número de dientes” [“rare congenital disease characterized by the absence of a number of teeth”].

Anotia
Anotia, the definition of which refers to the congenital absence of one or both ears, does not have entry in DRAE either.

Argyria
Argyria is a disease characterized by the change in skin color, from a natural color to a dark blue.

Blepharospasm
Blepharospasm is an abnormality in the function of the muscles of eyelids that causes unintentional closing of these. It is also associated with muscle spasms of the face.

Brachydactyly
The brachydactyly refers to an abnormal shortness of fingers or toes.

Cystinosis
As cystine appears defined, it would be consistent that cystinosis was reclaimed in the dictionary as a rare metabolic disease that causes accumulation of cystine in body tissues and thus the appearance of crystals of this amino acid in the cornea, marrow, lymph nodes and organs such as the kidney, mainly.

Cystinuria
The same is true concerning cystinuria, another uncommon, inherited disease that is characterized by a disruption of the reabsorption of basic amino acids that causes the formation of concretions in kidney, ureter and bladder.

Citrullinemia
Nor is there an entry for citrullinemia, metabolic disorder in which there is a deficit of Argininosuccinic acid synthase, an enzyme required for the incorporation of ammonia in the urea cycle.

Chondrosarcoma
Chondrosarcoma lacks an entry in DRAE, being a type of malignant bone tumor that develops in cartilage cells.
**Choroideremia**
Neither the choroid membrane nor the illness directly associated with it, choroideremia, are defined in the dictionary. This is a rare hereditary disease characterized by progressive degeneration of several layers of cells at the back of the eye.

**Cryptophtalmia**
The cryptophtalmia is the complete adhesion of the eyelids and is not collected in the *DRAE* either.

**Scaphocephaly**
There are no results for scaphocephaly in Academy’s dictionaries. It is another cranial deformity (as brachycephaly and plagiocephaly) that develops during the first months of life, affecting especially premature babies, and is characterized by the elongation and the narrowness of the head.

**Exencephaly**
Exencephaly is a malformation in which the brain is located outside the skull.

**Favism**
The lemma of favism refers to red cell related diseases.

**Fibrosarcoma**
The fibrosarcoma encompasses rare bone diseases or rare tumors.

**Fibromyalgia**
The medical term fibromyalgia, which refers to a set of musculoskeletal symptoms characterized by persistent pain and extreme fatigue, including psychological ones, has no entry in *DRAE*. However, words like acedia (sloth, slackness, but also sadness, anxiety) have a long lexicographical record in Academy’s dictionaries, since *Diccionario de autoridades* (RAE, 1726-1739), noting some of the clinical features that are often associated, for example, with fibromyalgia. The combination of physical and psychological disease components in the concept of acedia seems clearly linked with rare diseases like chronic fatigue syndrome or the aforementioned fibromyalgia.

**Galactosemia**
Galactosemia, a disease characterized by enzyme deficiencies that affect the metabolism of galactose, does not receive lexicographical treatment in dictionaries of the *Real Academia*, although they do have an entry dedicated to term galactose.
Gigantomastia
Gigantomastia is defined as a rare gynecological disease that involves excessive, diffuse and even disabling one or both breasts growth.

Glycogenosis
Glycogenosis, understood as a group of metabolic diseases characterized by a disruption in the process of formation and use of glycogen, is not included in Real Academia Española’s lexicographic repertoire either.

Hypochondroplasia
Unlike achondroplasia, the Academy’s dictionary does not include the term hypochondroplasia, referring in this case to the condition characterized by disproportionately short stature.

Hypertrichosis
Hypertrichosis is known as a congenital condition consisting in an increase both in the quantity and thickness of hair.

Hypopituitarism
The lemma hypopituitarism is not present in editions of DRAE. This is a medical term referring to an abnormal decrease in the hormones secreted by the pituitary gland.

Leukodystrophy
Genetic disorders grouped under the term leukodystrophy, resulting in the degeneration of fatty myelin sheath covering nerve fibers of the brain and adrenal glands, are also omitted.

Lymphangioleiomyomatosis
Lung ailment, the most common symptom of which is progressive respiratory dyspnea, known as lymphangioleiomyomatosis, is not defined.

Mannosidosis
Neither is mannosidosis, a hereditary disease caused by a disruption of carbohydrate metabolism that includes facial deformities and mental retardation.

Mastocytosis
Neither the term mastocytosis (abnormal growth of mast cells in the body), nor mastocyte (cell originating in stem cells from bone marrow and involved in mediating inflammatory processes) are included.
Nephronophthisis
There is no information in the dictionary on a condition called nephronophthisis, characterized by the small size of the kidneys with numerous small cysts.

Osteonecrosis
Osteonecrosis is a rare disease caused by decreased blood flow to the bones of the joints, entailing deterioration.

Pycnodysostosis
Pycnodysostosis is an unusual type of bone dysplasia.

Plagiocephaly
Plagiocephaly, already referred to in the section on scaphocephaly, is characterized by an asymmetrical distortion (lateral flattening) of the skull.

Retinoschisis
Retinoschisis, meanwhile, refers to the formation of intraretinal cyst, due to the separation of the layers of the retina.

Sialidosis
Furthermore, sialidosis is a metabolic disorder characterized by a defect in the sialidase enzyme. There is no defined term in the twenty-second edition of Academy’s dictionary for this condition.

Syringomyelia
These dictionaries do not reveal syringomyelia, a damage to the spinal cord due to formation of a fluid-filled area within it, either.

Discussion
After transferring the preceding data to percentage, a total of 37.03 % of the analyzed terms feature definitions in any of the various editions of DRAE, compared to 62.96 % of words lacking an entry in that glossary. Table 1 shows details of the date of incorporation of the terms selected on rare diseases that do appear collected in various editions of the Academy’s dictionary:
Table 1 – Dates of lemmatization of rare diseases

<table>
<thead>
<tr>
<th>YEAR OF PUBLICATION</th>
<th>PATHOLOGY</th>
</tr>
</thead>
<tbody>
<tr>
<td>1732</td>
<td>Dengue</td>
</tr>
<tr>
<td>1734</td>
<td>Leprosy</td>
</tr>
<tr>
<td>1884</td>
<td>Albinism</td>
</tr>
<tr>
<td></td>
<td>Diphtheria</td>
</tr>
<tr>
<td>1899</td>
<td>Brachycephaly</td>
</tr>
<tr>
<td>1925</td>
<td>Hydrocephalus</td>
</tr>
<tr>
<td>1927</td>
<td>Scleroderma</td>
</tr>
<tr>
<td></td>
<td>Ichthyosis</td>
</tr>
<tr>
<td>1936 [1939]</td>
<td>Acromegaly</td>
</tr>
<tr>
<td></td>
<td>Hemophilia</td>
</tr>
<tr>
<td></td>
<td>Microcephaly</td>
</tr>
<tr>
<td>1970</td>
<td>Achondroplasia</td>
</tr>
<tr>
<td></td>
<td>Botulism</td>
</tr>
<tr>
<td>1984</td>
<td>Thalassemia</td>
</tr>
<tr>
<td>1989</td>
<td>Brucellosis</td>
</tr>
<tr>
<td>2001</td>
<td>Phenylketonuria</td>
</tr>
<tr>
<td></td>
<td>Glioma</td>
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<tr>
<td></td>
<td>Legionellosis</td>
</tr>
<tr>
<td></td>
<td>Narcolepsy</td>
</tr>
<tr>
<td></td>
<td>Nevus</td>
</tr>
</tbody>
</table>

Source: Own elaboration.

When doing statistics according to the century in which these twenty words first appeared, the following graph is established:

Graph 1 – Distribution by century of the definitions of rare diseases

Source: Own elaboration.

As it can be seen in Graph 1, the highest rate of incorporation in the Academy’s dictionary of the terms for diseases of low prevalence selected to carry out this research took place in the first half of the twentieth century. The twenty-first century is left second place, taking into account that it has not advanced much and that DRAE only
has the 2001 edition and the progression of the twenty-third so far. The latter can be justified by the magnificent work of highlighting these conditions, along with the implied consequences, conducted by FEDER since the late twentieth century.

Having seen this history of absences, one might wonder what criteria the Real Academia applied to include some terms in the dictionary and not others, all of them relating to rare diseases. While it used literary sources from the beginning, it seems logical that the main reasoning to guide the inclusion of this medical terminology is a more or less frequent use of the terms by the speakers. But how to measure their use by an average speaker? Could it be a case of a not so objective judgment? In this regard, it seems significant that the DRAE does not incorporate fibromyalgia, a disease that has recently ceased to be regarded as less prevalent.

Perhaps these criteria, at first not very specific, became more evident when looking up entries grouped under the lemma syndrome or disease, where only those that seem to have a high social recognition and for which there is already public awareness are included. Incidentally, it seems advisable to include the syntagm rare disease in this second entry, or rather a less common disease as it has been called in recent years to avoid the metonymy in the connotations of rare; that is, to clear the mistaken belief that an individual suffering from a rare disease must be, by extension, odd.

This situation results from the goals initially set by the Diccionario de autoridades since it granted only secondary relevance to incorporating the technical vocabulary related to medicine, with reservation of creating a work dedicated solely to this question. Thus, only the more common or precise scientific terms were included, although there are examples that cast doubt to a successful completion of this objective. That is, the problem of the selection of entries is manifest from the first edition of this dictionary (GUTIÉRREZ RODILLA, 1993).

Clearly, the twenty-second edition is an improvement with respect to the lexicographical treatment of some of the specialized terms on low prevalence disorders. So much so that in this small selection, there are new lexicographic definitions for phenylketonuria, narcolepsy and nevus. The effort to adapt certain medical definitions to advances in science is also commendable, as in the cases of acromegaly or hydrocephalus.

However, there are still several entries that call for a reworking according to these scientific advances (eg incompletely specified symptoms of achondroplasia, or the definition of brachycephalic that is obscure to some extent, etc.). Moreover, the referrals of the definitions of certain nouns (as albinism, brachycephaly) to its corresponding adjective (albino, brachycephalic) are inconsistent when similar entries on nouns (eg, hydrocephalus) get a complete definition. Therefore, it seems advisable to remodel some of these lexicographical entries that have, in many cases, remained unchanged for several decades in the various editions of the Academy’s dictionary (for example brachycephalic or scleroderma).
As Gutiérrez Rodilla claims, the authors of the *Diccionario de autoridades* in the eighteenth century based themselves on the literary tradition to create a common vocabulary. Thus, the medical terms were included to help in the understanding of literary works, marginalising in that way the needs of language users in understanding the reality. “Es una situación que se continúa en gran medida en la lexicografía actual: no sólo en lo que ésta (sic) actitud tiene de razonable, sino también en sus rasgos más extremos, como el culto excesivo que nuestros diccionarios tienen al pasado [...]” (GUTIÉRREZ RODILLA, 1993, p.471).

From another viewpoint, without going into a detailed analysis on the formation of lexical units relating to rare diseases, both those collected in the *DRAE* and those that are not, it should be noted that they share the same morphological mechanisms as other more common words. For example, “aniridia”, “anodontia” or “anotia” (with the negative prefix “an-”), “exencephaly” (with the prefix “ex-”, meaning “out of”), “hypopituitarism” or “hypochondroplasia” (prefix “hypo-”, “under”), “hypertrichosis” (“hyper-”, or “above”).

Similarly, there are compounds as “hydrocephalus” (“hydro-” = “water” and “-cephalus” = “head”), “microcephaly” (“micro-” = “very small”) and “brachydactyly” or “brachycephalic” (“brachy-” gr. brachys – “short”, refers to “fingers” and “head”). Some words incorporate a second element denoting blood “-emia”: “galactosemia” < “galactose”; “citrullinemia” < “citrulline”; “chloroheremia” < “chloroid”; “thalassemia” < gr. thalassa, “sea”. The nominalisation with suffixes like “-osis” (“glycogenosis” < “glycogen”; “legionellosis” < “legionella”; “brucellosis” < “brucella”; “mastocytosis” < “mastocyte”; “sialidosis” < “sialidase”) and “-ism” (“albinism”, “botulism”) is frequent as well.

According to the above, it is not possible to determine if the cause of inclusion or exclusion of lemmas on rare diseases in Academy’s dictionaries, resides in an inconsistency with respect to the usual rules governing the processes of word formation in Spanish language.

**Conclusions**

1. This paper highlights the imprecision in the lexicographical criteria that manage the inclusion and treatment of specialized lexical units in Academy’s dictionaries; specifically, in terms to the area of medicine and health sciences, as accounted in the heterogeneous group of terms for rare diseases that have been selected. From a total of fifty-four conditions, only twenty (37.03 %) are lemmatized in *DRAE*. The absence of the term fibromyalgia is particularly noteworthy, since it has been considered a prevalent disease in recent years and yet, does not have an entry in the cited dictionary.

43 “It is a situation that persists largely in the current lexicography: not just inasmuch as this stand is reasonable, but also in its most extreme aspects such as excessive worship of our dictionaries for the past [...]” (GUTIÉRREZ RODILLA, 1993, p.471).
2. Consequently, it has not been possible, in this study, to deduce exact principles applied in the treatment of lexical entries on rare diseases. We, therefore, conclude that this group of terms does not appear systematically collected in Academy’s dictionaries, despite having the same characteristics as other words in general vocabulary.

3. The Academy’s dictionary satisfies the needs of the users (non-specialists, media professionals without formal training in medicine and health sciences, among others) seeking information on various lexical aspects (definitions, variations in the encoding of words, to name a few) of less prevalent diseases in a very partial manner. However, we must necessarily indicate the merit of the growing inclusion of terms related to these diseases in the different editions of DRAE, which has taken place since the last century to this day.

4. In any case, the dictionary of the Real Academia is a key reference, at least to any Spanish speaker, when resolving lexical-terminological doubts and, in this sense, one should not lose sight of its overall mission to clarify the meaning of those words, specialized or not, that are becoming part of the heritage of our language and being repeated daily in discourse, whether longstanding or not.

5. It would therefore be advisable that the dictionary extend the list of definitions for rare diseases, thus being able to generate an approximate and primarily informative, not scientific, idea of those considered more usual (for example, based on the index of references made to them in the mass media), in that way also expanding the general understanding of these conditions today.

6. It would also be expedient to undertake, from a scientific and specialized point of view, the arduous task of preparing a dictionary on rare diseases, or rather less frequent diseases, with the goal of making the terminology of these conditions more accessible to a nonspecialist public. While it is true that FEDER, on a national level, and EURORDIS, on European level, assemble in their online pages very useful and updated information on this issue.

7. A further step in the study of the presence of words that designate rare diseases in dictionaries should be consulting the dictionary of medical terms (Diccionario de términos médicos) from the Real Academia de Medicina (2012) and could be completed with the search in other directories as Vocabulario científico y técnico by the Real Academia de Ciencias Exactas, Físicas y Naturales (1996).


RESUMEN: Este artículo indaga en la trayectoria terminológica de cincuenta y cuatro enfermedades raras. Se analiza el grado de aceptación lexicográfica que tienen y han tenido –a lo largo del tiempo– en su tratamiento e incorporación en los diccionarios académicos de la lengua española. Hasta la fecha, tan solo veinte de los términos seleccionados están
lematizados en el DRAE, lo que representa el 37.03 %. Esta situación demuestra la inexistencia de criterios sólidos en la incorporación de términos nuevos sobre enfermedades raras en el citado diccionario general. El caso más reseñable es fibromialgia, patología que recientemente ha dejado de ser considerada una enfermedad poco frecuente y que, sin embargo, carece de lema en el DRAE. Si, por un lado, se debe asumir que el DRAE no es un diccionario médico especializado; por el otro, hay que valorar el grado de incorporación de estas voces en la vida social.


REFERENCES


Received in September 2014

Approved in October 2014