Maria Cristina Gatti /
Jeanette Hoffmann (eds.)

Storytelling as a Cultural Practice

Pedagogical and Linguistic Perspectives

PETER LANG

Storytelling as a cultural practice permeates all phases and areas of human life and opens up possible worlds. From their earliest days, children grow into a culture of storytelling, acquire language and literature, develop writing skills, and learn to communicate through storytelling in multimodal ways: orally and in writing, by playing, drawing, designing, singing, dancing and more. Through the process of narrating, experiences are structured, identities are formed, social contexts are shaped, and desires and futures are imagined. Narrative connects different times in history, various disciplinary fields in education and diverse linguistic-cultural spaces, but it also requires time and space itself. Against the background of an educational landscape that is currently competence-oriented, the question arises as to what role the art of storytelling plays in educational contexts, and what possibilities it opens up for learning. This edited volume aims to address this question, theoretically and empirically, from pedagogical and linguistic perspectives.

Maria Cristina Gatti (PhD) is Associate Professor of English Linguistics at the Free University of Bozen-Bolzano (Italy), where she also directs the Center for Academic Writing. She is an advisory member of the Center for Intercultural Dialogue (CID). As an applied linguist, she is currently leading research programmes on narrative in multilingual and multicultural settings, the role of English in multilingual education and professional environments, and language variation in transnational communication contexts.

Jeanette Hoffmann (PhD) is Full Professor of German Literature Didactics at the Free University of Bozen-Bolzano (Italy), where she also directs the EduSpace Children'sLiteratureLab. Previously, she was Full Professor at the Technische Universität Dresden and worked at the University of Education Upper Austria and the Freie Universität Berlin. Her research interests are graphic storytelling, reading engagement, literary learning and language education in multilingual and intercultural contexts.

www.peterlang.com

Maria Cristina Gatti / Jeanette Hoffmann (eds.)

Storytelling as a Cultural Practice

Pedagogical and Linguistic Perspectives

PETER LANG

Lausanne - Berlin - Bruxelles - Chennai - New York - Oxford

Library of Congress Cataloging-in-Publication Data
A CIP catalog record for this book has been applied for at the Library of Congress.

Bibliographic information published by the Deutsche Nationalbibliothek. The German National Library lists this publication in the German National Bibliography; detailed bibliographic data is available on the Internet at http://dnb.d-nb.de.

This publication was financially supported by the
Free University of Bozen-Bolzano

With the assistance of Martina Irsara, Cecilia Lazzeretti,
Mara Maya Victoria Leonardi and Elisabeth von Leon

Cover image: Giovanna Bampa

ISBN 978-3-0343-4505-7 (Print)
E-ISBN 978-3-0343-4833-1 (E-PDF)
E-ISBN 978-3-0343-4834-8 (EPUB)
DOI 10.3726/b21689

Published by:
Peter Lang Group AG, Lausanne, Switzerland

info@peterlang.com - www.peterlang.com

Contents

V Storytelling in specialised discourse

Kim Grego and Susanna Grego[1]

Narrative Medicine and Medical Narratives
Marfan Syndrome between Definition, Description and Narration

Abstract (English): This chapter examines the interconnection between narratives, descriptions and definitions in reports on Marfan syndrome. As a so-called rare disease, this syndrome is little known and may be difficult to recognize and diagnose. Following an interdisciplinary approach, the doctor's view on narrative medicine and the linguist's view on medical narratives are brought together. Four seminal reports on the condition are analysed, beginning in 1896, when the syndrome was described and defined for the first time. The analysis of the cases examines the presence of narrative elements in the description of Marfan's in the course of history and how these have affected the definitions of the syndrome. The cooperation between linguistic theory and medical practice is thought to be helpful in recognizing the condition, also potentially speeding up diagnoses.

Keywords: medical terminology, medical discourse, Marfan syndrome, Critical Discourse Studies, narrative medicine

Abstract (Italiano): Questo capitolo esamina l'interconnessione tra narrazioni, descrizioni e definizioni nei resoconti sulla sindrome di Marfan. Trattandosi di una cosiddetta malattia rara, tale sindrome è poco conosciuta, e può essere difficile da riconoscere e diagnosticare. Seguendo un approccio interdisciplinare, si associano il punto di vista del medico sulla medicina narrativa e il punto di vista del linguista sulle narrazioni mediche. Vengono analizzati quattro articoli seminali sulla malattia, a partire da quello che nel 1896 per primo descrisse e definì la sindrome. L'analisi dei casi esamina la presenza di elementi narrativi nella descrizione della sindrome di Marfan nel corso della storia, e come questi hanno influito sulle definizioni della sindrome. Si ritiene che la cooperazione tra teoria linguistica e pratica medica possa contribuire al riconoscimento della patologia, potenzialmente anche accelerando le diagnosi.

Keywords: terminologia medica, discorso medico, sindrome di Marfan, studi critici del discorso, medicina narrativa

---

1 Both authors are responsible for the ideas and research behind this study. Specifically, S. Grego authored 1.2, 3.3, 6.2 and K. Grego the remaining paragraphs.

1. Introduction

1.1 Narratives and medical humanities

Narratives found their official way in the clinical practice of medicine particularly since the late 20th century (cf. Salager-Meyer et al., 2013), and they have long and successfully been used ever since both by patients and by healthcare operators. Medical education has been looking at the inclusion of the history of the profession for the past 100 years, with recently renewed interest starting from "the professional turn toward 'social history' in the 1960s and 1970s" (Dolan, 2010, p. 393). Medical humanities, in turn, are an emerging interdiscipline that includes not only the history of medicine but also bioethics, narrative medicine, medicine in literature, creative writing, social sciences such as medical anthropology and sociology, as well as creative arts like music, painting, reader's theatre, and dance – in brief, "[a]nything that touches on 'the human condition', 'the humanising process' or 'the humanist philosophy' becomes relevant" (Dolan, 2010, p. 394).

Within this framework this chapter will firstly shortly introduce Marfan syndrome, then set out the research aims, illustrate the methodology, analyse the material and draw some conclusions.

1.2 Marfan syndrome

The condition considered here is Marfan syndrome (MFS), a so-called 'rare' disease (Orphanet, 2022a) which, for this very reason, is little known and may be difficult to diagnose. Marfan syndrome is "a systemic disease of connective tissue characterized by a variable combination of cardiovascular, musculo-skeletal, ophthalmic and pulmonary manifestations" (Orphanet, 2022b). When discussing a rare disease, it is useful to compare it to a better known one, so as to provide a term of comparison. Down syndrome (DS), for example, has a 1–5/10,000 prevalence and affects 1 in 400/3000 live births (with variations depending on the policies of individual countries) (Orphanet, 2022c). Marfan syndrome is found in 1 in 5000 individuals, and its prevalence is the same, estimated at 1–5/10,000 (Orphanet, 2022b).

One immediate consideration is, therefore, that one has about the same probability of *seeing* a person with Down's as one has the chance of seeing one with Marfan's. In reality, the difference lies precisely in the verb 'to see': while Down syndrome has almost invariably a very specific phenotypic manifestation (typical facial and body features), Marfan syndrome does so in some cases, and does not in others – and anyway the traits would on average be less

conspicuous. This is because the syndrome, first reported in 1896 by the French doctor Antoine Marfan, and caused by an only recently (1991) identified genetic mutation, manifests with a range of signs, especially cardiovascular, skeletal and ophthalmic ones. Of these, "[s]keletal involvement is often the first sign of the disease and can include dolichostenomelia (excessive length of extremities), large size, arachnodactyly, joint hypermobility, scoliotic deformations, acetabulum protrusion, thoracic deformity" (Orphanet, 2022b). Simplifying significantly, any very tall, lean and bent person with long fingers and eyeglasses may possibly be affected by Marfan syndrome or, of course, s/he may not.

Additionally, while individuals with Down syndrome often have cognitive impairments (which may result in noticeable behavioural expressions), those with Marfan are not, by definition, affected by intellectual disabilities. All this justifies the possibility that, while the probability of encountering a DS and a MFS person is the same, the perception of having done so might be completely different. This is reflected, for instance, by the fact that the US-based NORD, or National Organization for Rare Disorders, has Marfan Syndrome listed in its rare disease database, but not Down syndrome.[2] Two comparable genetic diseases, thus, may be treated differently, based not on their statistic prevalence but on the degree of awareness the public has of them.

A syndrome is defined as "a set of symptoms that occur together; the sum of signs of any morbid state; a symptom complex. In genetics, a pattern of multiple malformations thought to be pathogenetically related"[3] (*Dorland's Illustrated Medical Dictionary* 32nd edn, s.v. Syndrome). Like with other genetic syndromes, the (mis- / missed) diagnosis of MFS depends on descriptions: of signs or manifestations made by a patient, or of the patient's condition made by doctors in referrals. Indeed, the description of a disease, especially a syndrome, often takes the form of a narrative – especially in the past century and a half that has elapsed since it was first reported. It is argued that such descriptions can be equated to narratives or stories as understood in narrative medicine and that they play a key role for the definition and, therefore, the diagnosis of a disease. The relation between narrative description, definition and diagnosis will then be explored in the following sections, making specific reference to the case of Marfan syndrome.

---

2 As of January 31, 2024, Down Syndrome does not feature under 'D', while other trisomies appear under 'T'. https://rarediseases.org/for-patients-and-families/information-resources/rare-disease-information/

3 In the case of MFS, it is argued that it is best not to employ the term 'symptoms', which are subjective, but rather to talk about objectively observable 'signs' or 'manifestations'. This is the terminological choice that will be adopted throughout this study.

2. Aims and research questions

This study adopts an interdisciplinary approach, aiming at bringing together the doctor's view on narrative medicine and the linguist's view on medical narratives, by the example of Marfan syndrome. It insists, in particular, on the distinction between definition and description, a concept that may appear clear to the linguist but not to the practitioner, who focuses on clinical data to fit them into a given framework (Grego & Grego, 2021).

The research questions pursued in the section will be: 1) Could narratives be the connecting element, making sense of a sometimes subtle yet discriminating difference between the two concepts? 2) Does the presence of narratives affect the definition of the condition? 3) How much is narration present in descriptions of Marfan syndrome in past and recent times?

3. A mixed methodology

In order to answer these questions, a mixed methodology has been employed, drawing from linguistics as well as from the medical humanities. Reflections on text linguistics (De Beaugrande & Dressler, 1981) and specialized discourse (Garzone, 2020) have been combined with insights from the medical humanities (Dolan, 2010; Charon, 2006; Charon et al., 2017). A discursive interpretation of the social impact of the topic is offered as a conclusion, following Gouveia's (2003) understanding of discourse in Critical Discourse Analysis (CDA) as applied to a new science: "a probability pattern of a continual exchange of meaning in an inseparable web of relationships that include language, people, events, situations, institutions, social structures, and so on" (p. 55).

3.1 Description vs narration in text linguistics

De Beaugrande and Dressler (1981) in their influential work identify three functional typologies of texts: descriptive, narrative and argumentative. The latter of the three shall not be considered here (though the relevance of argumentation in medical texts is far from limited), so the focus will be on the first two categories. Descriptive texts are defined by the authors as being centred on objects or situations and based on frames as applied global patterns. Narrative texts, instead, focus on actions and events and the sequences in which they occur, and they mainly progress through schemas or "global patterns of events and states in ordered sequences linked by time proximity and causality" (De Beaugrande & Dressler, 1981, ch. IX, par. 6). Although De Beaugrande and Dressler (1981) provide quite clear-cut definitions, they are also the first

to comment that "Freedle – Hale (1979) found that narrative schemas can be acquired and transferred to the processing of descriptive ('expository') texts" (note 3), thus opening up to the possibility of mixing the various patterns. It is therefore admissible to hypothesize an alternation of descriptive and narrative patterns in medical narratives in which neither the mere description of facts nor the ordered narration of events stands alone, but they alternate, as with any real doctor-patient consultation.

3.2 Definitions in linguistics

In Western philosophy, definitions may be categorised according to various criteria, for instance their purpose. Longworth (2006) accordingly classifies them as stipulative (explaining meaning), abbreviative (favouring comprehensibility), explicative (approximating a meaning to another one) and descriptive or lexical definitions. The latter type, dedicated to descriptions, is the one of interest in this study and is thus defined:

> a sentence used to explain meaning. Less immediate purposes here include illuminating a less well understood *definiendum* by appeal to a better understood *definiens*; revealing the basis of one's understanding of the *definiendum*; or establishing dependence of the *definiendum* on the *definiens*. But the basic purpose of descriptive definition – explaining the meaning of the *definiendum* – is independent of the viability of these other purposes. This is good, since it would be surprising if many expressions in use were redundant (Longworth, 2006, p. 409, Italics by K.G. & S.G.).

3.3 Definitions in medicine

No such thorough definitions of 'definition' are given in medicine and the related fields; descriptive or lexical definitions are employed, and the notion is taken for granted. This notwithstanding, even medical lexicographers can be extremely accurate. For instance, in the current editions of the historical *Dorland's Medical Dictionary*, the Preface (Anderson, 2012) and the following Notes on the use of the dictionary report amply on how the entries have been constructed and organized linguistically, mentioning syllabication, pronunciation, cross-referencing, official terminology, proper names and even eponyms. However, one singular aspect that clearly emerges is the dependence of definitions on descriptions: "[t]he entries are enhanced by 1525 illustrations, nearly all in full color, each chosen for its ability to clarify the *description* given in the *definition*" (Anderson, 2012, p. vii, Italics by K.G. & S.G.). It is argued here that, in spite of the medical focus, this statement is intrinsically linguistic in nature, and that reference is made to a type of text as in De Beaugrande and Dressler

(1981): the encyclopaedic entry. It is furthermore suggested that the overlapping of the two concepts – description and definition – should be considered a key issue in conditions like Marfan syndrome, which are expressed with a range of different manifestations that vary individually and cannot thus be limited to a univocal 'description'. If a medical condition is *diagnosed* based on a definition, and if it is *defined* based on a description, then it may be diagnosed based on a *description*. An erroneous, partial or changed description may therefore result in a physician mis-diagnosing a patient. The notions of definition, description and narration debated in this section come together and are given enhanced significance from the perspective of narrative medicine.

3.4 Narrative medicine

Narrative medicine is especially centred on stories and storytelling as powerful tools of diagnostic investigation, for reporting and establishing and maintaining effective doctor-patient relationships. According to Zaharias (2018), writing from the perspective of the family physician,

> [s]tories are our life's blood. We like to listen to stories, and it is through stories that we make sense of the world, that identity is shaped, and that we attempt to communicate what matters to us. This is well recognized by psychology, the social sciences, and the humanities, where narrative ideas originated. (p. 177)

It is significant that this notion has currently reached even the primary level of healthcare in society (family doctors, General Practitioners, family physicians or any other name they go by), as the result of a journey toward awareness and the recognition of the complexity of the human condition. It has not always been so: indeed, it is thanks to scholars like Rita Charon that narrative medicine has been established in North America, initially, and then elsewhere in the world, especially in English-speaking and Western countries. In the volume co-edited by Charon in 2002, several authors started a reflection around narrative, ethics, stories *and* medicine. From the sequencing of the human genome (in process at the time, completed now) seen as an unfolding story of its own, or the voices behind consent forms, to be hermeneutically interpreted as in a Pirandello play (Churchill, 2002), to psychiatry consultations conceived as an attempt by patients to escape social narratives imposed upon them (Martinez, 2002), the acknowledgement of an underlying connection between how illness is *told*, how it is *interpreted* and what comes out of it in diagnostic and therapeutic terms emerged as pivotal for doing medicine. Subsequent work by Charon (2006) refined the notion of narrative medicine, now defined as

> a unifying designation to signify a clinical practice informed by the theory and practice of reading, writing, telling, and receiving of stories. The name appealed to me because, as a nominal phrase, it points to a 'thing' and not an idea (fulfilling William Carlos Williams's dictum that there are no ideas but in things) and connotes a kind of practice along with a set of conceptual relations in which it nests (p. viii).

The idea of 'narrative competence' is also introduced, i.e. the fact that, once the awareness of the relevance of the narrative has been assimilated, the ability to use such competence capably, both as a patient and as a practitioner, may "attain that illuminated grasp of another's experience that provides them with diagnostic accuracy and therapeutic direction" (Charon, 2006, p. 11). Four fundamental doctor-patient divides, or chasms separating the sick and the well, are then laid out: the idea of death ('relation to mortality'), the condition of illness ('contexts of illness'), the reasons behind diseases ('beliefs about disease causality'), and the negative emotions linked to illness ('shame, blame, and fear') (Charon, 2006, pp. 22–24). These are identified as perspectives under which the doctor-patient relation runs the risk of collapsing, because they tend to be seen as routine by experienced practitioners, but always cause wonder and sufferance with patients: addressing them with the help of narratives thus becomes the scope of this new approach.

Finally, in *The Principles and Practice of Narrative Medicine* (Charon et al., 2017), narrative medicine having firmly established itself, the didactic aspect is explored, and suggestions are put forward as to how to integrate ethics, philosophy and literature in medicine through the common lens of narration. The times having changed too, the bioethics discourse is also tackled and often referred to, with the concept of 'close reading /writing' emerging as the key tool to *do* narrative medicine. Indeed, the rationale behind the 'close' premodification is as simple as the other ideas informing the approach: what is needed is attention, since "[b]oth a model for and an avenue toward attention, close reading fortified with attention to its subjective dimensions has become narrative medicine's laboratory and training ground" (Charon et al., 2017, p. 158).

The story of narrative medicine has been told chronologically here, in a Western-style past-to-present modality. However, the authors believe that the innovative approach it introduced may also be applied, in historical terms, to a time and a story that occurred before it was even conceived – or, at least, this is what the next sections will attempt to do.

4. Material

For the purpose of this study, four seminal studies on Marfan syndrome were analysed, beginning with the very first one that contributed to describing and defining the syndrome:

T1. Marfan, A. B. (1896). Un cas de déformation congénitale des quatre membres plus prononcée aux extrémités caractérisée par l'allongement des os avec un certain dégrée d'amincissement. *Bulletins et memoires de la Societe medicale des hopitaux de Paris 13*, 220–226.[4]

T2. McKusick, V. A. (1955). The cardiovascular aspects of Marfan's syndrome: a heritable disorder of connective tissue. *Circulation 11*(3), 321–342.

T3. Dietz, H. C. et al. (1991). Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. *Nature 352*(6333), 337–339.

T4. Loeys, B. L. et al. (2010). The revised Ghent nosology for the Marfan syndrome. *Journal of Medical Genetics 47*(7), 476–485.

5. Analyses: medical narratives of MFS

5.1 Marfan (1896)

The six-page article that Antoine Marfan presented to the Société médicale des hôpitaux de Paris in 1896 is, like several other seminal research papers[5], a short and almost unassuming report that has nonetheless become a milestone in its field and, terminologically speaking, has come to name a condition that would only be explained as the result of a genetic mutation two centuries later. The use of eponyms in medical terminology is a fashion that has come and gone, and has been discussed at length, including by one of the authors with respect to the very same syndrome (cf. Vicentini et al., 2016). The fact that Marfan's name was given to the condition he described is thus not at all unusual for the time, nor is the accurate and lengthy description of his 'patient 0'. What is argued here is that, rather than a description proper, i.e. a factual report of an

4 "A case of congenital deformation of the four limbs, more pronounced at the extremities, characterized by elongation of the bones with some degree of thinning". Perhaps a superfluous clarification, this is a French text. Being a seminal paper, it is usually quoted in the original in the literature. Although a translation into English could easily be attempted, also in this case using the original was deemed both acceptable and preferable.

5 Cf. e.g. Watson & Crick (1953) on the structure of DNA, or Wilmut et al. (1997) on cloning Dolly the sheep.

object or a situation, it includes narrative elements like actions and events, told in sequential order.

It is a common expectation that a medical report should be objective, partial and detached; yet, just like a patient is not merely an 'object', the report may take on a narrative tone that turns it into a story-like creation, though based on observed facts. Marfan (1896) starts out with what could be considered a powerful incipit, regardless of the textual genre:

[01] *Gabrielle P.*… est âgée aujourd'hui de cinq ans et demi. ("*Gabrielle P.*… is currently aged five and a half", T1: 220, Italics by K.G. & S.G.)

The observed object is a five-year-old girl, known by her first name and by only the initial letter of her surname. Although at the time this was not surprising, nowadays she would have been called by her initials, G.P., or assigned a number, e.g. P1 for Patient 1. However, back then like in the present day, naming a character has always had the biblical potential to infuse life into it: this is precisely what happens with Gabrielle. Indeed, if one thinks of Franz Kafka's *Trial*, written only some 20 years later, it is evident that its protagonist, Joseph K., is attributed a first name and only the initial of his surname as a mimic of a professional report in a specialised setting – a lawsuit brought before a judge – but it could well have been a doctor describing the condition of a patient – as in Dr Marfan and Gabrielle P. A black-and-white medical photograph completes the picture and gives the girl a face as well as a name.

Lexically, reporting verbs (cf. [02]) appear in the medical description, as other stories are told within the main one:

[02] Pendant que la mère était grosse de notre petite malade, **elle raconte que** dans le premier mois, elle fut fortement impressionnée par la vue d'un homme brûlé. ("While she was pregnant with our little patient, the mother **relates** that in the first month she was greatly impressed by the sight of a burnt man", T1: 221, bold type by K.G. & S.G.)

Their presence, of course, initiates forms of indirect discourse (Toolan, 1988[2001], pp. 116–118), which in turn imply, among other features, hypotactic syntactic relations and deictic indicators (cf. [03]) keyed to the perspective of the narrator, who becomes the undisputed master-of-puppets in the construction of the narrative:

[03] **Quand nous avons reçu l'enfant**, il y a bientôt trois ans, elle **était âgée** de deux ans et demi […]. ("**When we were referred the child**, almost three years ago, **she was** two and a half years old", T1: 225, bold type by K.G. & S.G.)

There are also instances of indirect thought:

[04] […] **nous avons pensé** qu'elle était et qu'elle resterait idiote ("**we thought that** she was and would remain an idiot", T1: 225, bold type by K.G. & S.G.).

This clause, apart from matching Toolan's (1988[2001]) view that "Indirect Thought is 'normal' as the means for reporting anyone else's thoughts besides your own, including a character's in a heterodiegetic narrative" (p. 138), serves various purposes: it is the narrator's own indirect thought; it has the speech-act power of a medical opinion (in that it is, after all, the opinion of a doctor); but at the same time it is also an autobiographical narration in which the *passé composé* tense signals an action in the past that is relevant at the time of the narration, also in the past and, semantically, creates expectation as to a change of mind or a development. Indeed, this is precisely what immediately follows:

[05] **Nous nous sommes trompés**; son intelligence **s'est développée** tardivement, mais parfaitement, même dans le **pauvre** milieu d'une salle d'hôpital ("**We were wrong**; her intelligence **developed** late but perfectly, even in the **poor** environment of a hospital ward", T1: 225, bold type by K.G. & S.G.).

The medical description – supposedly of an object – is no longer such: it has become a narration, in which the protagonist's story intertwines with the narrator's own story, who is also no longer just a detached observer. In turn, the little patient has become a subject, a person, in whose fortunes the scientist has empathetically become involved: the reader may suspect it from the phrase "*tardivement*, mais *parfaitment*" ("*late*, but *perfectly*") – which juxtaposes a negative fact and a positive one in a theme-rheme sequence, thus insisting on the positive – but is sure of it when the author expresses evaluation in "*pauvre* milieu" ("*poor* environment"). The medical opinion was wrong, and the doctor is not unhappy about it – there might even be a hint of pride in his authorial voice. The *petite fille* has become a full character, Gabrielle P., whose author is Antoine M., a clinician who has learned to care about his little patient and expresses pity for the 'poor' environment in which she had to grow up. This is precisely what Charon et al. (2017) call for when they express "our commitments to relationships in patient-centered care and our conviction that narrative competence can widen the clinical gaze to include personal and social elements of patients' lives vital to the tasks of healing." (p. 1)

5.2 McKusick (1955)

The second text to be examined is almost 60 years younger than Marfan's. Authored by a renowned geneticist, who studied genetic syndromes, including among the genetically famed Amish population, it shows evidence of all the changes and developments occurred in the medical field over that time. To begin with, it is much more structured compared to T1. Twenty-two pages long, it includes a short abstract, a description of what was known about the disease until then, with a classification and explanation of its main cardiovascular aspects, then it reports information about 50 families "in which at least one *bona fide* instance of this condition has occurred" (T2, p. 321). The clinical reflections are interspersed with reports on specific cases, in detail six individuals and one "unusually severely affected family" (T2: 337).

Terminologically, it is worth signalling that the condition is referred to in slightly different ways, all referring to Dr Marfan (of course, this did not apply to T1):

- Marfan's Syndrome;
- Marfan's syndrome;
- Marfan syndrome;
- Marfan's disease;
- Marfan mutation;
- Marfan trait;
- abnormality of the Marfan type.

The variation in the use of capitalization and of the possessive 's attests to an uncertainty in the terminology, meaning that at that time in history the notion that multiple apparatuses and organs were involved and multiple signs may or may not be present (a 'syndrome') was already clear. Conversely, a tradition of considering or calling it a single individual 'disease' could possibly still be predominant. An alternation of 'syndrome' and 'disease' used synonymously for euphonic reasons could be hypothesized, although they do not always necessarily alternate in the test.

It is the seven specific stories of individuals and families, though, that are mostly of interest in terms of description and narration. The first datum is the naming of subjects:

[06] L.K. (J.H.H. 571745), a white man born in 1913 (T2, p. 326),
[07] R.L.L., a 37 year old unmarried white man (T2: 329),
[08] B.J.P. (H.L.H. A93754) was born Nov. 9, 1951 (T2: 330),
[09] M.E.C. (J.H.H. A98174), born in 1940 (T2: 331),

[10] M.E.R. (J.H.H. 176836), a colored female born in 1929 (T2: 333),
[11] K.B., a 24 year old white man […] (T2: 335),
[12] Individual I-1 died suddenly in 1897 at the age of 47 years […]
The son of this man (II-5) died at the age of 27 years […]
individual II-6, an intelligent observer and cooperative informant […]
Her husband (III-5) died in 1945 at the age of 32 years […]
The daughter of this man (IV-7) was born May 17, 1937. […]
The brother of this girl (IV-8) was born Nov. 27, 1938. […] (T2: 337).

No longer having a first name, they are now identified by their initials only, and sometimes by a code (J.H.H. most likely being the Johns Hopkins Hospital, where McKusick worked for decades). This choice is of course in line with the usage of more recent times, which definitely protects the patients' anonymity, although it limits them to being exclusively that – patients. The presence of photographs of various subjects, clearly showing their faces, may be used to challenge the notion of anonymity. Yet, the images are clinically detached in style and it can be argued that the audience of the American Heart Association's journal was a strictly professional one – in this regard, there is no difference with the way Gabrielle P. is also shown to the public, in T1.

Expectedly, the lexicon of the article is also more impersonal with respect to Marfan's report of 1896, if for some informal choices, like the verb in

[07b] R.L.L., a 37 year old unmarried white man, **dropped dead** on March 19, 1953 (T2: 329, bold type by K.G. & S.G.),

that would perhaps not be deemed suitable in the present day, when great care is put into using respectful expressions regarding patients. At the same time, the figurative power of the phrase contributes to the creation of a story, in what may sound, for instance, like an effective news article lead.

The narrative features must then be sought at the syntactic level, especially in the clause-sentence constructions. Thus, moving further into the story of R.L.L.,

[07c] R.L.L., a 37 year old unmarried white man, dropped dead on March 19, 1953, **while being interviewed** for employment (T2: 329, bold type by K.G. & S.G.),

one discovers in the temporal subordinate clause that the punctual action of dying occurred while a continuous action was ongoing, "being interviewed". This, on the one hand, creates a chronological sequence or a schema (De Beaugrande & Dressler, 1981); on the other hand, it introduces information that is

relevant not only for the medical *subject* but also for the *person*. Details such as R.L.L. being unmarried or that he was in the midst of a job interview when he suddenly died are significant for the clinician because, when heart conditions are discussed, the marital status or the stress of the competitive situation are known to have an influence on them. Contextually, the very same details build on the narrative, in this case with a satellite providing circumstantial information: e.g., 'during a stressful event' would have offered the expert the same knowledge but not the same *story*. The numerous temporal syntactic elements make useful examples of the presence of schemas in the paper. One such element is represented by temporal subordinate clauses, as in [07c], or in

[13] the patient was relatively well until April, 1952, **when she had a first attack** of paroxysmal tachycardia (T2: 331, bold type by K.G. & S.G.).

Another one is expressed by the tenses; a past perfect creates a complex timeline in the past,

[14] The mother **had had** no pregnancies in the 19 years between this one and that which occurred in 1933 (T2: 330, bold type by K.G. & S.G.).

A present perfect used to express duration provides a temporal connection with the present, which makes the story more relevant and possibly timely again:

[10b] M.E.R. (J.H.H. 176836), a colored female born in 1929, is a member of a family which **has been known** to this hospital for about 25 years (T2: 333, bold type by K.G. & S.G).

To sum it up, although the style of T2 reflects the changed times and tends, accordingly, toward lexical impersonality, the narration still clearly emerges from the syntactic level of the construction of the story. This is interesting in that, while vocabulary may be chosen consciously, the crafting of words into syntactic structures may be more of an unconscious act when performed by the scientific writer, unless, of course, such a writer also has the human story at heart, as may be in the case of a doctor. McKusick does not perhaps build as powerful a story as Marfan does with Gabrielle, but the ethical concern is clearly expressed in his writing:

[15] This kinship illustrates one of the difficulties of genetic research in man. The **illegitimacy of this infant** and the presence of a legitimate wife made the **utmost tact** and **resourcefulness** necessary for collecting even these few data. The father of the infant is about 74 inches tall, has long hands and feet, and wears spectacles. Examination was not possible and no further pedigree information was obtained.

Whether this only reflects the scientist's greed for data or the doctor's apprehension for the difficult social condition of the newborn patient B.J.P. (cf. [08]), whose "heart was extremely overactive and **shook the whole bed**" (the quantitative datum is missing, but the picture is clear, T2: 330) and died "at the age of **only** eight months" (the dates of birth and death are given, readers can make their own calculations, T2: 331) is not for us to say.

5.3 Dietz et al. (1991)

If T1 gave a name to the syndrome and T2 made its familial nature evident, T3, yet another seminal paper in the history of Marfan's, first provided an explanation for the condition. The time gap between T2 and T3 is 36 years, but there is a narrative link between McKusick, one of the first supporters of the mapping of the human genome, and the team, also mostly based at Johns Hopkins: Harry C. Dietz is the Victor A. McKusick Professor of Genetics at Johns Hopkins University School of Medicine who identified a mutation in the *FBN1* gene as the cause of the very same syndrome that inspired McKusick to pursue medical genetics.

T3 was published in 1991 as a letter to *Nature* and, much like Watson and Crick (1953, cf. note 8), it reports a milestone discovery in just about two-and-a-half pages. Synthesis is thus the underlying feature of this specific text and of the genre itself in the past 40 years or so (Garzone, 2020, pp. 138–140). T3 indeed shows all the characteristics of the contemporary scientific research article, including extreme nominalization, depersonalization, synthetization and schematization (Garzone, 2020). The years separating T1 from T2 may be more than those between T2 and T3, but the technological developments occurred between 1955 and 1991 make the distance between the texts much more relevant. This is reflected first and foremost by the terminology, providing names for entities, concepts and procedures that did not exist before, and which shows all the typicalities of specialized lexicon, e.g. monoreferentiality, technicality, artificiality (Garzone, 2020, pp. 32–34). 'Marfan syndrome' – without the possessive 's and a lowercase 's', in line with the preference for synthesis – has now become lexicalized as the term used to identify the condition and is used consistently throughout, cf. [15] and [16]. The absence of the possessive 's may also represent a form of depersonalisation, in that the syndrome no longer 'belongs' to Marfan. In particular, the technicality at the lexical level and the depersonalization at the syntactic one emerges as creating a feeling of detachment between author and text, and between author and receiver. This is confirmed by the (obviously) short descriptions of the two patients mentioned in the article.

[16] This patient (E.S.) had **severe** manifestations of the **Marfan syndrome** noted in early infancy including increased body length, arachnodactyly, muscular hypotonia, joint laxity and mitral regurgitation. Subsequent complications included pectus excavatum, scoliosis, high-grade myopia, lens dislocation and severe cardio-vascular pathology necessitating mitral valve replacement and repair of ascending and descending aortic dissections. She **died as consequence of congestive heart failure** at **age 17**. Her parents and brother are **clinically well**, without signs of the Marfan syndrome except for isolated mitral valve prolapse in the brother, a finding seen in up to 8 % of the general population (T3: 338, bold type by K.G. & S.G.).

The first patient [16] is identified by her initials, E.S., and her age is only mentioned in regard to her death, which occurred at 17: anonymity is maximized, characterization minimized. Her death appears different, for example, from R.L.L.'s (cf. [07]) – who theatrically "dropped dead" in front of his potential employer – aseptically caused by a three-word term saying what failed and not what actively killed her. Her age at the time of death, which could universally be agreed on as untimely, is not evaluated, unlike in B.J.P.'s case, who died at "*only* 8 months". Ironically, though, the rest of her family are said to be *well*, but the specification is provided that they are only well "clinically", to make sure the family's wellness is not wrongly attributed to either their psychological or emotional state. The second patient, B.C., receives even less space in the article.

[17] The patient (B.C.) was diagnosed **in infancy** on the basis of arachnodactyly, joint hypermobility and ectopia lentis. Now aged 19 years, she has developed **the additional** manifestations of long-bone overgrowth, severe scoliosis, foot and leg deformities, mitral and tricuspid valve prolapse and regurgitation and severe dilation of the aortic root. Neither parent has any features of the **Marfan syndrome**. **Paternity was confirmed** as previously described for patient E.S. (T3: 338, bold type by K.G. & S.G.).

Her age is given at the time of writing, and her story only unfolds by mentioning her infancy, when she was first diagnosed, and the present time, when she shows further manifestations, defined "*the* additional", with the definite article coldly pointing to the full range of Marfan manifestations. Anonymity is guaranteed, so there is no ethical conundrum in stating that paternity, for both patients, was confirmed – the very datum whose absence so frustrated McKusick in his description of B.J.P. (cf. [08]). No photographs appear of the two patients – rightfully *subjects*, here – where the only images are from

single-stranded conformation polymorphism (SSCP) analyses: the focus has shifted from the phenotype, with the patients' long limbs and big myopic eyes, to the genotype, from the macro to the micro level of genetics.

Far from criticizing T3 for its impersonality, it is clear that it is the result of a stylistic change affecting all scientific-academic written genres in those decades (cf. Garzone, 2020, especially pp. 105–107). Indeed, the magnitude of the discovery itself and the complexity of the technology behind it is such that, even within the medical community, its communication has become restricted to a certain number of hyper-specialized experts who are able to grasp it in its entirety. Synthesis is thus paramount, since everything else about the syndrome must already be known to them and is considered background information, not to be focused on. The attention is all for the genetic-level details, of which the patients have become mere vessels, at most providing extra information against which to check the micro-level hypotheses. As cold as this may seem, the big picture is that such fine analyses, in the past 30 years, have returned invaluable data for the early diagnosis and treatment of many Marfan syndrome patients that, if asked, could certainly tell happier stories than E.S. or B.C.

5.4 Loeys et al. (2010)

T4 represents the end of this Marfan syndrome story, seen historically as one unfolding discursive narration. The work of a large panel of international experts, from both American and European top research institutes, it reviewed and adjusted a previous nosology (De Paepe et al., 1996), in turn correcting the so-called 'Berlin nosology' (Beighton et al., 1986). Already fourteen years old at the present time, it has not so far been replaced by newer, definitive versions. The main innovation it brought forth was a stronger focus on the cardiovascular manifestations; it also determined that "aortic root aneurysm and *ectopia lentis* are the cardinal clinical features […] [and] *FBN1* testing, although not mandatory, has greater weight in the diagnostic assessment" (T4: 476). The shift toward genetic testing represents, in a way, the final destination of a journey that started out with the classic observation of phenotypes and manifestations. At the same time, the testing not being compulsory and renewed attention being placed on vascular disease, it also represents a step back to clinical evaluation – in a sense, back to the patient. Since they depend on technological developments and on the subsequent progress of research resulting from evaluating significant numbers of cases, trends in medical approaches are subject to adjustments over time, so further revised nosologies of MFS may ensue. From the perspective of the narrative descriptions adopted here, what is interesting

about T4 is not so much the guidelines and detailed specifications that make up the bulk of it but the initial paragraph. No individual patients' stories are reported, but the entire history of the syndrome is briefly summarized:

[18] **Since Antoine-Bernard Marfan described** the **5-year-old Gabrielle** with skeletal manifestations of the disease that now bears his name, important progress has been made in the delineation of the **Marfan syndrome** (MFS) and recognition of associated risks. The main features of this autosomal dominant disorder include disproportionate long bone overgrowth, ectopia lentis and aortic root aneurysm. In 1955, **Victor McKusick** first established a classification of connective tissue disorders, which resulted in the publication of his monograph 'Heritable connective tissue disorders'. In 1986, an **international panel of experts** defined a set of clinical criteria (**Berlin nosology**) for the diagnosis of MFS […]. Following the **identification of *FBN1*** (encoding fibrillin-1) as the causal gene for MFS, it was recognised that the Berlin criteria falsely allowed a diagnosis of MFS in individuals with a positive family history of MFS, who had only non-specific connective tissue findings themselves and who did not carry the mutation present in more typically affected family members. New diagnostic criteria were therefore put forth in 1996, referred to as the **Ghent nosology** (T4: 476, bold type by K.G. & S.G.).

As in T3, 'Marfan syndrome', with its abbreviation, is the established specialized term being employed. The narration begins with a temporal subordinate clause introduced by 'since', providing a sense of incipit: it is the equivalent of the proverbial 'there once was a' – doctor, in this case, whose name was Marfan. Indeed, of the characters of this short story, three have a first name (Antoine-Bernard, Gabrielle, Victor); two of these also have surnames (Marfan, McKusick); one is a collective noun, "an international panel of experts"; the last they avoid mentioning, making recourse to a depersonalization strategy, i.e. agent-less passivization ("criteria were therefore put forth"), which is typical of specialized texts (Garzone, 2020, p. 105). It is significant that both Marfan and McKusick have acquired mythical status in the narration: they are the only MFS experts quoted by name and actually, at that, by name and surname – while the 'Surname (year)' citation style typical of academic writing is set aside in their honour. Indeed, nobody else is mentioned between 1896 and 1955, confirming the direct narrative link between the two physician-characters. It should also be noted that McKusick was also among the authors of the Berlin nosology, and that he died just two years before T4 was published. Conversely, the panel behind the Ghent nosology included several experts that also authored T4, as

well as Harry C. Dietz, first author of T3 and also behind "the identification of *FBN1*"; hence, the depersonalization could also serve hedging purposes (Garzone, 2020, p. 119). Gabrielle, conversely, only defined by her name and very young age, has lost the initial of her surname, which also induces a mythization process: with her surname's initial she was an anonymized patient, with just her first name she becomes a fictional character – like a Caesar or a Napoleon – who anyone dealing with Marfan syndrome knows and need not define any further. The paragraph thus takes on a celebratory tone, as at a party where the main protagonists of modern-day research on Marfan's meet, with the significant exception of Gabrielle, the only patient and involuntary heroine of the story. T4, in its introductory paragraph, stands out as a hybrid text, comprising a description with narrative features, personal and even celebratory mentions of characters from the past, depersonalization and hedging strategies, as well as hyperspecialized terminology typical of domain-specific texts towards the end, when dealing with more recent and contemporary actors and events.

6. Conclusions

The research questions this chapter set out to explore regarded 1) whether narratives could be identified as connecting the concepts of description and definition, 2) whether the presence of narratives affects the definition of the condition and 3) how much narration is present in descriptions of Marfan syndrome in past and recent times.

6.1 Narrative descriptions: an inseparable bond

The four texts analysed – in spite of their differing age, styles, sub-genres within medical literature writing – all seem to include forms of narration that are part of the descriptions they feature. To this end, narratives may indeed be considered a unifying element. In particular, from the linguistic perspective, descriptions appear to blend smoothly with narratives. Indeed, the cases examined show how narratives in/of Marfan's syndrome decrease since its 'discovery' (i.e. its first attested description), being closer to storytelling in earlier times (1896 and 1955), diminishing to a minimum in the 1990s when reporting the genetic nature thereof, but again gaining momentum in the revision of the MFS nosology, which starts out as a pure and celebratory narration in its review of the literature.

6.2 Narrative definitions: medical ambiguity

From the medical perspective, definitions stemming from narrative descriptions, which could be re-termed 'narrative definitions', emerge as potentially ambiguous, and easily mistakable for definitions, especially in the decades leading to the familiar first and then genetic explanation of the condition. For example, if physicians working between 1896 and the 1950s or even later looked for exactly the same description of Gabrielle P. to diagnose Marfan's – i.e. considering the definition of the syndrome inseparably attached to its first description(s) – they would have missed, as is likely, numerous cases. In other words, Marfan syndrome has, in the course of history, had the sets of variable signs characterizing it been taken as *definitory* and not as *descriptive*, hence delaying any innovative, prompt recognition of new cases.

6.3 Medical narrative descriptions as discourse

Narratives are present in medical descriptions and their relevance for description-based definitions and diagnoses highlights the role of the narrator in the discursive construction that s/he necessarily provides in his/her storytelling. When narration occurs in contemporary science, as in the case of Marfan syndrome, only slightly older than a century in its attestation, we ought to be reminded of how the 20th century "fragmentation and specialization of disciplines has turned the scientist into a specialized ignoramus" (Gouveia, 2003, p. 55). If the science at stake is medicine, though, the scientist cannot afford to be one such specialized ignoramus, but should constantly be aware of all the human and social implications of a doctor's role – whether it is the lab-based geneticist or the family physician examining a Gabrielle-like patient. For this reason, the interdisciplinary approach adopted for this study, according to Gouveia (2003), can help reduce the discipline fragmentation that has been affecting new science, particularly in the last three or four decades of the 20th century, if we see it not as regarding disciplines but *themes*: "[a]s opposed to discipline fragmentation, thematic fragmentation involves the migration of local concepts and ways of reading to other contexts in a localized but total and interconnected description. In other words, thematic fragmentation involves *operationalization*" (Gouveia, 2003, p. 55, original Italics).

Thematic fragmentation would thus allow researchers, from any field, to address the same themes from various methodological perspectives in an operative or practical way, thus not separating the validity of science "from its usefulness and utility which will help to define its internal consistency and cogency" (Gouveia, 2003, p. 48). Applied to the case of Marfan syndrome,

specifically, and how it has been described historically, if "nothing in discourse has meaning as an isolated entity but can only be understood as interconnections between various processes of observation and measurement" (Gouveia, 2003, p. 56), then the cooperation between medical and linguistic observation is thought to be helpful, potentially, to speed up diagnoses by an interdisciplinary understanding of 'definition', 'description' and 'narration'.

Primary sources

Marfan, A. B. (1896). Un cas de déformation congénitale des quatre membres plus prononcée aux extrémités caractérisée par l'allongement des os avec un certain dégrée d'amincissement. *Bulletins et memoires de la Societe medicale des hopitaux de Paris*, *13*, 220–226.

McKusick, V. A. (1955). The cardiovascular aspects of Marfan's syndrome: a heritable disorder of connective tissue. *Circulation*, *11*(3), 321–342.

Dietz, H. C., Cutting, G. R., Pyeritz, R. E., Maslen, C. L., Sakai, L. Y., Corson, G. M., Puffenberger, E. G., Hamosh, A., Nanthakumar, E. J., & Curristin, S. M. (1991). Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. *Nature*, *352*(6333), 337–339. https://doi.org/10.1038/352337a0

Loeys, B. L., Dietz, H. C., Braverman, A. C., Callewaert, B. L., De Backer, J., Devereux, R. B., Hilhorst-Hofstee, Y., Jondeau, G., Faivre, L., Milewicz, D. M., Pyeritz, R. E., Sponseller, P. D., Wordsworth, P., & De Paepe, A. M. (2010). The revised Ghent nosology for the Marfan syndrome. *Journal of Medical Genetics*, *47*(7), 476–485. https://doi.org/10.1136/jmg.2009.072785

References

Anderson, D. M. (2012). Preface. In W. A. N. Dorland (Ed.), *Dorland's Illustrated Medical Dictionary* (32nd ed.) (p. vii). Saunders.

Beighton, P., de Paepe, A., Danks, D., Finidori, G., Gedde-Dahl, T., Goodman, R., Hall, J. G., Hollister, D. W., Horton, W., & McKusick, V. A. (1988). International Nosology of Heritable Disorders of Connective Tissue, Berlin, 1986. *American journal of medical genetics*, *29*(3), 581–594. https://doi.org/10.1002/ajmg.1320290316

Charon, R. (2006). *Narrative Medicine*. Oxford University Press.

Charon, R., DasGupta, S., Hermann, N., Irvine, C., Marcus, E. R., Rivera Colón, E., Spencer, D., & Spiegel, M. (2017). *The Principles and Practice of Narrative Medicine*. Oxford University Press.

Charon, R., & Montello, M. (2002). *Stories Matter: The Role of Narrative in Medical Ethics*. Routledge.

Churchill, L. R. (2002). Narrative ethics, gene stories, and the hermeneutics of consent forms. In R. Charon & M. M. Montello, *Stories Matter: The Role of Narrative in Medical Ethics* (pp. 187–199). Routledge.

Beaugrande, R. D., & Dressler, W. U. (1981). *Introduction to Text Linguistics*. Longman Publishing Group.

De Paepe, A., Devereux, R. B., Dietz, H. C., Hennekam, R. C. M., & Pyeritz, R. E. (1996). Revised diagnostic criteria for the Marfan syndrome. *American Journal of Medical Genetics*, *4*, 417–426. https://doi.org/10.1002/(sici)1096-8628(19960424)62:4<417::aid-ajmg15>3.0.co;2-r

Dolan, B. (2010). History, Medical Humanities and Medical Education. *Social History of Medicine*, *2*, 393–405. https://doi.org/10.1093/shm/hkq005

Dorland, W. A. N. (2012). *Dorland's Illustrated Medical Dictionary* (32nd ed.). Saunders.

Freedle, R. O. & Hale, G. A. (1979). Acquisition of new comprehension schemata for expository prose by transfer of a narrative schema. In R. O. Freedle (Ed.), *New Directions in Discourse Processing* (pp. 121–135). Greenwood.

Garzone, G. (2020). *Specialized Communication and Popularization in English*. Carocci.

Gouveia, C.A.M. (2003). Critical discourse analysis and the development of the new science. In G. Weiss & R. Wodak (Eds.), *Critical Discourse Analysis: Theory and Interdisciplinarity* (pp. 47–62). Palgrave.

Grego, S. & Grego, K. (2021, June 28–30). *Accuracy, definition, integrity in medical terminology*. [Conference presentation]. The 19th International and Interdisciplinary Conference on Communication, Medicine and Ethics (COMET), University of Insubria, Como, Italy.

Longworth, G. (2006). Definitions: Uses and Varieties of. In K. Brown (Ed.), *Encyclopedia of Language & Linguistics* (2nd ed.) (pp. 409–412). Elsevier.

Martinez, R. (2002). Narrative understanding and methods in psychiatry and behavioral health. In R. Charon & M. M. Montello (Eds.), *Stories Matter: The Role of Narrative in Medical Ethics* (pp. 129–140). Routledge.

Orphanet (2022a). *About rare diseases*. Orphanet. https://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN

Orphanet (2022b). *Marfan syndrome*. Orphanet. https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=558&lng=EN.Orphanet

Orphanet (2022c). Down syndrome. Orphanet. https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=870&lng=EN

Salager-Meyer, F., Alcaraz Ariza, M. Á. & Luzardo Briceño, M. (2013). The Medical Narrative from a Diachronic Perspective (1840–2009): Titling Practices and Authorship. In M. Gotti & C. Sancho Guinda (Eds.), *Narratives in Academic and Professional Genres* (pp. 293–318). Peter Lang.

Toolan, M. (1988/2001). *Narrative. A critical linguistic introduction*. Routledge.

Vicentini, A., Grego, K., & Canziani, T. (2016). A matter of terminology, when terminology matters: Naming common genetic diseases. In G. E. Garzone, D. Heaney & G. Riboni (Eds.), *Language for Specific Purposes* (pp. 200–218). Cambridge Scholars Publishing.

Watson, J. D., & Crick, F. H. C. (1953). Molecular Structure of Nucleic Acids: A Structure for Deoxyribose Nucleic Acid. *Nature*, *4356*, 737–738. https://doi.org/10.1038/171737a0.

Wilmut, I., Schnieke, A. E., McWhir, J., Kind, A. J., & Campbell, K. H. S. (1997). Viable offspring derived from fetal and adult mammalian cells. *Nature*, *6619*, 810–813. https://doi.org/10.1038/385810a0.

Zaharias G. (2018). What is narrative-based medicine? Narrative-based medicine 1. *Canadian family physician Medecin de famille canadien*, *64*(3), 176–180.