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On the indiciary nature of the clinical method: an anthropological view of a published case study

Sobre el carácter indiciario del método clínico: una mirada antropológica a partir de un relato de caso publicado

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ABSTRACT In this article we present a published case study as an object of reflection. On this basis, we carried out a partial reconstruction of the process of study and diagnostic elaboration of the Uruguay syndrome, showing the circumstances of the case, the selection and interpretation of "clues," and some of the details relevant to the clinical reasoning. Our starting point is the recognition of the narrative nature of clinical knowledge and of the clinical method as an indiciary method. The manuscript of the Uruguay syndrome has a narrative structure adjusted to the conventions of a scientific article, which gives lesser importance to the clinical method. We carried out diverse methodical encounters, mainly involving in-depth interviews with the authors of the manuscript and observation in their workplace. The text seeks to recover the histories of work based on the indiciary or semiotic model of knowledge, and recognize the importance of this model in medical practice.

KEY WORDS Anthropology; Clinical Medicine; Medical Genetics.

RESUMEN En este artículo presentamos un relato de caso publicado como objeto de reflexión, sobre el que se realizó una reconstrucción parcial del proceso de estudio y una elaboración diagnóstica del síndrome Uruguay, mostrando las circunstancias del caso, la selección e interpretaciones de "pistas", y algunos de los detalles que fueron relevantes en el raciocinio clínico. Nuestro punto de partida es el reconocimiento del carácter narrativo del conocimiento clínico y del método clínico como un método indiciario. El manuscrito del síndrome Uruguay presenta una estructura narrativa ajustada a las convenciones del artículo científico que pone al método clínico en un segundo plano. Nuestros encuentros metódicos fueron diversos y comprendieron, sobre todo, entrevistas en profundidad a los autores del manuscrito y observaciones en su lugar de trabajo. El texto propone recuperar las historias de trabajo basadas en un modelo de conocimiento indiciario o semiótico y reconocer su importancia en la práctica médica.

PALABRAS CLAVES Antropología; Genética Médica; Medicina Clínica.

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INTRODUCTION

During 2000, the *American Journal of Medical Genetics* published an article describing a new genetic disease: the Uruguay facio-cardio-musculo-skeletal syndrome (1) (a). The syndrome is defined as an autosomal recessive disease linked to the X chromosome which presents facial, muscular, skeletal and cardiac alternations. This article focused on the semiological characterization of the syndrome (2,3), describing, from the clinical point of view, four members of the same family.

This manuscript (b) was written by clinical physicians from genetics institutes in both Montevideo and Los Angeles (California), and came to us through one of its authors (Andrea Quadrelli's father), who is a clinical geneticist. Therefore, we became aware of this publication and the study itself in a family context, through accompanying (in the sense of sharing information and different experiences) part of the research process.

When we first read the published manuscript we were extremely surprised, because we could not recognize or understand the information presented. In this article we offer some of the findings of the study we developed as part of our doctoral thesis which essentially constitutes an attempt to comprehend why the analyzed article was so strange and uncomfortable to us. In our work, we attribute an anthropological motivation as well as personal circumstances (c) to the fact the Uruguay syndrome manuscript was our principal object of study.

Our starting point is acknowledging the narrative nature of clinical medicine (5). The daily practice of a clinical doctor is full of stories (especially, those "opening" stories patients narrate to physicians), and the medical discourse itself regarding disease assumes a story format (6). In this way, the case description – a central element in the education of medical students and in clinical practice – is probably the most illustrative example of a conventional narrative in a medical environment.

The case report or "medical case" involves a narrative rationality which is developed in daily medical practice (5,6). This narrative rationality can be compared to the work of a detective who, when faced with a particular set of circumstances (the case), adopts a rational procedure defined as abductive (d), interpretative, retrospective and narrative (6). In other words, the physician performs circular reasoning, going from the effects to the causes, which begins with the analysis of signs and symptoms. The physician has to compare signs in a context in which often their expression is ambiguous and may hold different meanings. Despite this, the physician has to choose one of the possible meanings by performing an interpretative or abductive process to create a hypothesis about the possible causes until he or she can come to a viable conclusion or preliminary diagnosis. In this sense, clinical reasoning is characterized by a circular movement between generalization (biological abstractions) and particularization (the individual patient).Therefore, medical knowledge is built through practice, although not at the expense of scientific information, creating hypotheses/abductions which, articulated with inductive/deductive reasoning, lead to the elaboration of the diagnosis (8).

Thus, it is important to acknowledge medicine as a practice based in the diagnostic skills and clinical experiences (e) of physicians, because, when faced with a patient, the physician can only become aware of a disease indirectly through interpreting the signs s/he observes and the symptoms described by the patient (5). For this reason, physicians work in conditions of "inescapable uncertainty" (6 p.4) which demand practical reasoning or *phronesis* which acquires – according to Aristotle – a clear definition in anthropological and moral terrain (9). That is to say that *phronesis* implies not only the practical sense or flexible interpretative capacity that allows for determining the best way to proceed according to the circumstances in order to achieve certain goals, but also, as Gadamer highlights, "the sense for setting goals themselves and taking responsibility for them" (9 p.62). This interpretative capacity is one of the principal characteristics of clinical reasoning whose distinctive methodology, medical semiology, consists of the interpretation of the signs and symptoms of each individual patient to build a retrospective chronology of the disease (f). In this way, the research method of medical semiology, the clinical method, depends not only on the technical or scientific knowledge of the doctor, but also on his or her personal or professional experience.

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Defining clinical medicine as an interpretative practice (10,11) facilitates its articulation with the circumstantial or semiotic model of the historian Carlo Ginzburg (12). He sustains that "though reality may seem to be opaque" (12 p.177) there are privileged zones, signs or clues, which make deciphering it possible. In this way, clues (g) are presented as privileged tools to understand the realities which are difficult to apprehend, through the analysis of singular and specific expressions with broader dimensions.

Ginzburg defines medicine as a circumstantial discipline, essentially qualitative, the objects of which are cases and individual situations (12). Therefore, the reason for the lack of certainties in medicine derives from the indirect and circumstantial knowledge of the disease (the living intangible body) and the fact that for each individual the disease can adopt different characteristics. For this reason, the narrative is one of the principal ways to represent a disease and its causes, because it leaves space for circumstantial aspects and for the "contingency, conjuncture, and multiplicative causes that unfold over time" (6 p.80).

At this point, it is important to acknowledge the existence of at least two different narratives: the story the patient presents to the physician and the story later reformulated by the physician. As Montgomery explains, the medical narrative may be strange or incomprehensible to the patient, because, in the medical translation or reinterpretation, the patient's own existence is modified or almost erased. Indeed, the medical narrative can be considered "alien" with respect to the patient's version (5 p.13)

In this work, and after reading the manuscript of the Uruguay syndrome, it is also possible to affirm that we experienced the published narrative as "alien." This "alien" form was the main source of our discomfort or astonishment (h) which fundamentally stems from a critical attitude (13) – that is to say, a philosophical-anthropological attitude which places our beliefs on hold so that we can question ourselves about its causes and meanings. Thus, this paper can be inscribed within a perspective of medical hermeneutics (9) mainly defined by the effort to comprehend and the "call for a coherent and responsible proposal towards the construction of more inclusive and productive interactions in the matters of health" (14 p.559).

The manuscript of the Uruguay syndrome

The article under study offers a type of narrative which reports a rare case (i): the story of medical research carried out by geneticists regarding an unexpected disease. In the manuscript (1), the narrative construction of this case takes its most condensed and reformulated form in pursuit of a strict narrative pattern that, among other functions, tries to control those aspects deemed subjective, that is, not only does it limit the experience of the patient to the reconstruction of the events from a medical perspective, but it also presents a passive narrative voice and a standardized narrator (6,15,16). As an example, the paper utilizes the undifferentiated compound author-narrator-observer (6), despite the fact that the article has more than one author and more than one physician was involved in the study of the family. Thus, the writing style reflects the differences between providing care to patients and the elaboration of the diagnosis itself.

We previously acknowledged that the medical narrative has an alien form that, when translated by the physician, returns to the patient in such a way that their own experience is unrecognizable in the new narrative. We also revealed that we ourselves experienced the published written narrative as alien, thus our initial surprise. Nevertheless, Montgomery (5) explains that the alien form of the medical narrative is caused by the distortion and flattening of the patient's experience. In the "case" of the Uruguay syndrome manuscript, what experiences or issues might be excluded?

Our first reading of the manuscript puzzled us because something was missing: the *objective* perspective of the published narrative had overshadowed the clinical, circumstantial method itself, characteristic of medical semiology and the basis for clinical reasoning. Missing were the circumstances of the case, the selection and the interpretation of "clues" and the operative and critical details needed for the diagnosis in the interpretative, circular reasoning of physicians.

The medical narrative organizes the medical history of the patient as part of a "diagnostic journey" (17), made up of the interpretative reconstructions of the information gathered by the physician. The patient's words and body are analyzed for clues and signs that may allow the physician

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to learn more about the experience they represent (6). However, the physician must make sense of the patient's history in conditions generally accepted in the "scientifically-oriented world" (6 p.124) through a case report or an *objective*, explanatory article with scientific status.

Regarding the manuscript, a long time had passed between the writing of the case report and our reading of it, and even more time had passed since the process of studying the family (j). Nevertheless, we took the risk of recovering the "entanglement" of the diagnosis (5,6) – that is, the narrative reconstruction providing an explanation of the sequence of events, the signs of which were analyzed and interpreted by geneticists – or of recovering, as Ginzburg (12) would say, stories of circumstantial work.

MATERIALS AND METHODS

In order to provide a context for the published article, we used different approaches and held a number of methodological meetings. The results described in this paper arise mainly from in-depth interviews (18) carried out by Andrea Quadrelli with geneticists, the authors of the manuscript. Throughout 2012, fifteen interviews were carried out, which were recorded and later transcribed to facilitate their analysis from a qualitative perspective. The confidentiality of the interviewees was protected, therefore different letters will be used in this article to identify the quotes corresponding to each physician. In addition to the interviews, other information was gathered using the technique of participant observation, the objective of which is, traditionally, to recognize situations in which social and cultural universes are expressed and generated in their full articulation and variety. These observations were carried out during workdays of approximately 8 hours each, over a period of three months at the Institute of Genetic Medicine (IGM) [*Instituto de Genética Médica*] in Montevideo. Briefly, the IGM is made up of six doctors (geneticists, gynecologists, obstetricians and pediatricians), laboratory personnel (approximately 10 staff members who are biologists or chemists) and administrative personnel. The IGM offers the *services* of genetic counseling, prenatal and postnatal diagnoses of genetic and multifactorial diseases, molecular cancer studies and filiation tests. Our analysis attempts to adopt an ethnographic way of seeing (19,20) in the sense of undertaking a descriptive effort so as to develop a cultural interpretation. Appropriate informed consent was requested of all participants. The results shown in this paper are part of our doctoral thesis: *"O manuscrito da síndrome Uruguai debaixo da lupa: olhares sobre os médicos clínicos geneticistas a partir de um relato de caso publicado"* which was approved by the ethics of investigation committee of Escola Nacional de Saúde Pública, Fundação Oswaldo Cruz (CAAE: 01585312.4.0000.5240).

RESULTS: PRESENTATION AND ANALYSIS

The presentation of the Uruguay syndrome as an "autosomal recessive disease linked to the X chromosome" defines the disease as a genetic, monogenic, Mendelian condition, with an inheritance pattern consistent with a recessive disorder linked to the X chromosome.

Diseases are a central category of medical knowledge. However, this and other concepts characteristic of the medical rationality are relegated to the "terrain of the implicit" (2 p.50) and can be only inferred from a study of discourse and medical practice (21). Camargo Jr. identifies a "generic frame of discursive construction" (2 p.57) surrounding the elements of the category of disease made up of three principal axes: the explicative, the morphological and the semiotic.

The explicative amounts to the characterization of disease as a process, this being the tenet of physiopathology, in which medical knowledge is closer to the "hard sciences," under the domain of biology. The morphological is related to the description of typical harms, which is the basis of pathological anatomy, in which all the devices used to perform supplementary examinations are included. Finally, the semiotic corresponds to the reading of cases, to clinical knowledge itself, where diseases are considered to be "constellations of signs and symptoms, creating semiotic gestalts" (2 p.59).

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The objective of diagnosis is the characterization of diseases in the most complete way possible, detailing the harms present and their evolution over time, ideally identifying the original causes (etiology) (k). Laín Entralgo (23) highlights two meanings central to the term "diagnosis": diagnosis as a result or a clinical judgment, and diagnosis as a process or an art. One of the authors of the manuscript (M) expresses this distinction in the following way:

> *I remember Rodríguez, a famous surgeon. I was a young man of 21, 22 in the university hospital* [...] *I can see him now* [...] *they called him at night for an emergency consult, and Rodríguez says, "this is appendicitis." And the intern doctor of the emergency room asked him: "Why is it appendicitis?" And he answered, "It smells like appendicitis." Ok, that's it, that's telling you that his diagnosis is based on a series of associations acquired over his years of experience. And it was indeed appendicitis, I mean, "it smells like appendicitis," that's not a scientific affirmation, but it's a medical affirmation, because it has a scientific basis, the patient's pain and a series of associations made, it's the art of medicine. It's a practice based on science but where there's also a lot of art.*

The expression "it smells like appendicitis" is related to one of the most known medical aphorisms: "clinical practice is king." In these cases, it is used to refer to the physician who has acquired clinical knowledge or practical wisdom or *phronesis* (6,11,24) which is enough to recognize that "smell." In fact, the apparent paradox between art and science establishes a tension in medicine itself where, in most cases, art refers to the relatively subjective skills of physical diagnosis, or more precisely, a tacit knowledge: "the hunches that experienced physicians have without quite knowing how" (6 p.30). Montgomery considers that the science-art duality obscures the practical nature of medicine which includes both clinical experience and information of a scientific nature, where clinical judgment or phronesis is highlighted as an essential quality which allows physicians to adapt their knowledge to the experiences and circumstances of each patient (6).

One of the ways to understand the tension inherent in the science-art duality is to view the affirmation "medicine is an art" from a rhetorical perspective of a metaphorical nature. That is to say, "medicine is *like* an art" in relation to the intuitive nature of certain clinical skills. By so doing, it is reasonable to consider the clinical-diagnostic-therapeutic process as a whole which implies a combination of inductive, deductive and abductive reasoning (where art forms part) which may vary on a case-by-case basis.

Medical semiology is defined as the treatment of clinical examination methods (3) which involve the investigation of signs and symptoms, the search for physiopathological explanations, and the critical analysis of the collected data for the formulation of diagnostic hypotheses. It is an essentially practical activity performed in direct contact with the patient and the starting point of the clinical experience (10). In a schematic division, it could be said that the anamnesis deals with the symptoms while the physical examination involves the signs (l). Physicians interpret and build their own medical narrative using the stories within the patient's' narratives (anamnesis) and the observation and recording of the signs of the disease in their bodies (physical examination) (6,11). Both stages represent the reconstruction of a profile which corresponds to a semiology and location within the general schema of disease, carried out through clinical practice (2,17).

"We performed a semiological diagnosis"

The manuscript of the Uruguay syndrome primarily corresponds to the semiological axis responding to clinical knowledge, related to information that comes directly from the examination of the patient. The physicians thus define the published article as a clinical, radiological and genealogical description. In this way, the manuscript presents a typical semiologic *gestalt* which defines a new disease through the identification of a group of signs and symptoms. L and M explain:

> *It begins with a clinical event, absolutely clinical, because it is based on a genealogical event, a semiological event, given by the descriptions of malformations. And we*

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> *established a nosological diagnosis, the diagnosis of a new entity, not yet defined, and with a cause we know to be genetic, because of the genealogical events, above all* [...] *Apart from the nosological diagnosis* [...] *we made a semiological diagnosis using clinical events, specific malformations or deformities, and certain radiological information.*

The identification of "malformations" or "deformities" is possible by comparing them to an ideal "normality," this relation being one of the characteristics of the medical rationality. This relation was well characterized by Canguilhem (25) who recognized that without the concepts of normal and pathological, the thoughts and actions of physicians would be incomprehensible.

On the other hand, the following explanation from physicians L, S and M regarding the study of the family arises from the definition of disease as a *process,* which we recognized above as being part of the explicative axis, holding physiopathology as its model discipline and *cause* as a central category:

> *We have already described this family, but we have not studied them from a physiological, and especially physiopathological, point of view yet. We still do not have a definitive molecular result.*
> *The family has not been completely studied...*
> *The basic research part of the disease we weren't able to carry out due to financial reasons, that's where basic research, science, physiopathology comes in, the physiopathological definition of the Uruguay syndrome is basic research, it's not part of art, the art of the physician lies in the diagnosis* [...] *It's something which that has not been described yet, and is important not for the fact of describing it for the first time but also because through that you can learn about normal physiology. Physiopathology helps you to understand normal physiology.*

Clinical geneticists link the explicative and the semiological axes by placing the manuscript in the diagnostic process. In this process, they insist, the medical *art* is discovered, referring to a "sensory knowledge" (22 p.133). In other words, the individual with Uruguay syndrome, the case, is the principal category of the semiological axis, which presents clinical medicine as its model discipline, hence the reference to art or "medicine as an art" because, in this axis, the method is principally circumstantial and involves, as mentioned above, a combination between reasoning and the definition of a disease as a semiological *gestalt*.

On the other hand, within the clinical doctor-patient encounter, the physiopathological mechanisms do not play an important role beyond the clues that they can offer in relation to the patient's story, as the doctor's appointment is focused, mainly, on understanding the meaning of the symptoms and resolving them somehow (6). The manuscript of the Uruguay syndrome reflects this perspective, its principal objective being to define a diagnosis. Physiopathological aspects will be set aside for another moment, for another article. One possible explanation is related to the concept of medicine as a practice and the ethics of that practice. The need to treat the patient's disease will tend reduce the issue of the cause to the minimum manifestation of symptoms possible (6).

"It is an article with strict research methods"

This statement belongs to N, one of the authors of the manuscript. As explained above, a semiotic approach, such as the model of medical semiotics, is based on the interpretation of clues or signs (12). In that model, a new specific work method is recognized. An analysis of the diagnostic process of the Uruguay syndrome, according to the circumstantial model, implies the acknowledgement of a particular method of work and knowledge production, since the diagnosis is not explicit for the physician; on the contrary, it is the result of a joint narrative elaborated between the doctor and the patient (17). Therefore, stories related to the diagnosis are expressed as something reached, presenting the diagnostic process as a "journey" (17 p.96). Our research sought to uncover that journey.

The study of the family affected by the Uruguay syndrome began with a medical consultation two years before the manuscript was

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published, on the part of a 19-year-old male who we will call Juan, sent to IGM "*for diagnosis*" by orthopedic surgeon P.

P, director of a traumatology and orthopedic ward of a general hospital in Montevideo, had known Juan since he was a child. According to L, Juan's medical history had several stages: first, a problem with his hip, then, a problem with his spine and, lastly, a problem with his hands and feet. P stated:

> *They had an excellent description from the professor, and he, as an experienced individual, had no diagnosis. When the traumatologist examined* [Juan] *during adolescence and with his medical history* [...] *they began to treat his spine, but when he developed the condition in his hands and feet during adolescence, well, one could only think, using basic judgment, that it must be linked to a single cause. That is when he was referred to the genetics department.*

In the first testimony, L directly refers to the "life experiences" (6) of P, which is an essential component of the clinical experience. In the second testimony he describes the development of Juan's disease, including an unexpected and complicated hip dislocation, years later malformation of his spine and, some time later, alterations in his hands and feet. We can see here the conception of disease not as a static object but as a narrative that develops contingently over time (6). In this sense, physicians must make sense not only of the signs and symptoms but also of the progression of the disease. More time is necessary as well as more clues and other interpretations, which denote the importance of time and the context of clinical perception and its interpretations. Regarding this aspect, L explains:

> *We don't always keep searching. It's often happened that 10 years elapsed between examining a patient for the first time and establishing a diagnosis, because the diagnosis is the result of the evolution of the disease. New phenotypical, clinical or paraclinical elements emerged that made the diagnosis possible.*

As we highlighted previously, the elaboration of a hypothesis using a group of clues or signs based on a physical examination of the patient or the semiologic study implies a certain quotient of guesswork or abductive induction (26). The observations made by P, who were used by L, may be considered a form of abduction, of logical inference. During the interview, P stated that the moment he saw the patient with "*those large malformations*" in his hands and feet, he thought: "*this has a first and last name*" and immediately referred the patient to a clinical geneticist.

In this paper, we propose a characterization of clinical geneticists as semiotic consultants, a type of Sherlock Holmes, as described by Ginzburg (12,27) and Montgomery (5,6). In the diagnostic journey of the Uruguay syndrome, geneticists may be presented as restless researchers/hunters of clues, tracks or signals.

After Juan's first consultation at IGM, L remembers: "*we evaluated the condition and we did not have a diagnosis for his deformity.*" Consequently, the physicians began to study the patient according to the precepts of medical semiology (anamnesis, physical examination, additional tests). A series of malformations or anomalies were identified, without achieving a diagnosis.

L also explains that "*within the methodology of genetic study,*" once the patient has been examined, it is necessary to examine their parents and relatives. As L and M explain,

> *…in order to see if any of them had any minor manifestation, in order to orient ourselves, as is commonly done. We had already requested to examine his parents and his two sisters.*
> *The clinical event refers to the patient but, in the case of genetics, it refers to the patient and family or just the family, because the objective of genetics is not the patient, but the family. In regular medicine, you talk to the patient and the diagnosis is for that patient, the diagnosis of cholelith is for the patient. But in genetics, the patient is the family, the objective is the family, which means there is a clinical practice regarding the family, creating a good genealogy, how what is observed, the trait, the feature, the symptom and the sign, is passed down…*

For a clinical geneticist, the family medical history is important because it is the key to the

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diagnosis, in order to prove that a certain disease is hereditary, to offer information regarding its evolution and manifestations and to define a heredity pattern (28). Maybe, for this reason, the first figure of the manuscript shows a family tree that includes 178 individuals distributed through seven generations (m).

Most of Juan's relatives lived over 500 kilometers from Montevideo and their contact with the physicians was delayed. However María, one of Juan's aunts on his mother side, who was 25 years old and worked as a housecleaner in Montevideo, was informed of the studies requested by the doctors regarding their family by her sister (Juan's mother). Therefore, four months after Juan's first consultation, María approached IGM. As L explains:

> *She came and said, "I'm worried because I have brothers, cousins and uncles who have malformations in their hands and feet"* [...] *She described her worries, her concerns about the possibility of having children with the problems she had seen, not so much because of her nephew, but because of her experience with her uncles.* [...] *Because she knew that some had died and the family remembered that Juan was very much like the other relatives who died young due to heart conditions, in addition to their crippling malformations.*

María traveled periodically to Rivera, therefore she became the main connection to Juan's family. Several times, María submitted different sources of information to IGM such as pictures of Juan's childhood in which, according to L, "*some of his deficient motor skills could be seen.*" Additionally, Juan's sisters and maternal grandparents traveled to Montevideo for María's wedding and visited IGM.

As part of a "*work methodology,*" physicians carry out consultations with other physicians or institutions when faced with patients whose diagnosis cannot be "determined." Regarding the participation of a US physician specialized in bone dysplasia in the study of this family, L remembers:

> *We contacted him because, due to our work methods at the institute, we are constantly writing to one place or another for consultations* [...] *and he responded with something that was very useful. He said he was married to an Argentine woman, that he was planning to travel to Buenos Aires in order to deliver lectures and that he could visit us if we so desired* [...] *It was a fortuitous situation because he was contacted for a consultation and he offered to come. For us, it was wonderful.*

A year after Juan's first consultation, a meeting at the orthopedic ward of a public hospital in Montevideo was organized in order to present the undiagnosed patient cases to this specialized physician. When Juan's case was presented, L recalls that the US physician said, "'*I don't know what it is. I don't know what problem he has' and he seemed very interested.*" Immediately, L wrote in Juan's medical history: "*This is something new.*"

The physician's confirmation of the lack of similar cases, following a process described by Montgomery as "narrative argumentation" (5 p.45), encouraged the geneticists at IGM to study the family. In this initiative, María played a central role. L comments:

> [María] *was the driving force who helped us, who brought the family together, who coordinated the interviews with the great aunts. I remember drinking tea with several of these women* [pointing to the family tree] *here in Montevideo, at least three times. I held several interviews and* [María] *always came with me.*

Additionally, the medical team decided to travel to the north of the country to meet the rest of the family. Once again, María went with the doctors on this first trip and coordinated a meeting with most of the family members in a single house. About 20 people attended the meeting. L and M remember:

> *She came with us* [...] *to take blood, to examine everybody clinically and to take pictures* [...] *She had coordinated everything.*
> *By the time we arrived at the house everyone was there, there were a ton of people. We were there for three hours and we examined everybody's hands and feet. We saw who was affected and we visited them again later*

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> *so that we could record information in a calmer setting.* [...] *We explained that due to the number of cases in the family, it was highly likely that we were facing a genetic disease that needed to be studied. We were there to do that, to see who was sick or had problems.*

After visiting this first house, the team visited two more, which, according to L, were *"short and social visits"* and a third house where a first cousin of María lived. He was one of the men affected by the syndrome described in the manuscript. Again, María led the physicians to this family by coordinating and acting as intermediary in the relationship with them.

On this trip, three men with the disease were identified and a consult was scheduled with them at the hospital. A short time later, on a second trip, the physicians met "the affected males" and Juan's mother at the hospital. M comments:

> *Later we took another trip, but we met them at the hospital. There we examined all the men that were affected, there were three. We saw them again, in a calmer setting, and we took pictures and x-rays.*

On this second trip, once again with María, the physicians interviewed María's aunt, who had provided care and attention to her two deceased brothers, both of whom had apparently had the disease. Despite the fact that just months earlier the woman had thrown away their medical test results, the physicians managed to obtain a picture of one of her brothers from an identity document that she still kept. This picture is published within the manuscript.

On this trip, with María as well, the physicians visited another house where Juan's great-great-grandmother, aged 98, lived in order to gather data on the family tree. L and M recall:

> *We then visited the elderly lady, the eldest of them all, who gave us additional family information, here she is* [pointing at the family tree]. *She was over 90 years old and she gave us lots of information about the family* [...] *She was very clear and she gave us accurate information about dates and years. She remembered perfectly all of the deceased.*
>
> *When you are trying to make a family tree, you find the eldest family member, who is the one who knows the most.* [María] *knew this woman was alive, so we went there.*

During the process of studying the family, geneticists are found to be semiotic consultants, both in the search for and identification of signs, clues or symptoms. After Juan's first consultation, the physicians acknowledged their radical lack of knowledge: "*I don't know what it is,*" which led them to develop multiple strategies in order to determine a diagnosis, employing skills with which to arrive at a clinical understanding. Indeed, clinical reasoning is more apparent when there is an unknown, multiplicative and non-linear diagnosis (6). Recalling some details about the study of this family, L describes:

> *I went there, to the orthopedics department, with a recorder and sat with the head of the orthopedic and trauma ward so that he could talk to me about and and interpret the x-rays for me, so he could say: "I see this and that" and explain to me the most important features that should be highlighted as pathological, the least normal features. He gave me the radiological description, I recorded everything and I later transcribed it.*

This testimony, which describes part of the research strategies that were carried out, is interesting because it brings to light the exercise of distinguishing the normal from the pathological. It also shows the research procedure: "*So that he could…interpret the x-rays for me.*" In other words, analyzing x-rays also requires specialized and circumstantial knowledge.

The participation of the US physician, himself considered to be an experienced semiologist, is a clear example of the search or hunt for the signs in the journey of diagnosis for the Uruguay syndrome. In the previously mentioned presentation of patients without a diagnosis, L affirms:

> *I remember the first patient he saw came because of his short stature. The doctor asked the patient to take off his clothes, saw his "shawl" scrotum and diagnosed him with Aarskog syndrome. We had never diagnosed*

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someone with that syndrome and everybody was shocked.

The specialized physician, like Sherlock Holmes, skillfully diagnosed a patient through a series of minuscule perceptions which constituted a hypothesis.

María's participation, a sort of key informant in anthropological terms, shows a constant concern for recovering clues.

In summary, physicians identify signs and symptoms to construct primary hypotheses tested through anamnesis, the patient's physical evaluation and additional tests. Similar to the reasoning of a detective trying to solve a crime, the diagnostic hypothesis is constructed and reformulated through practical and interpretative reasoning (5,6,11). The reconstruction of both a crime and the narrative of a diagnosis convey stories of circumstantial work that are important to understand the case.

The name *Uruguay*

The manuscript presents a diagnostic result (23) that makes reference to clinical judgment: the medical meaning, or what the physician recognizes in a patient. The diagnostic result presented in the narrative record of the manuscript does not specify the circumstantial method of the diagnostic process and it also obscures the contingency and context of the research carried out, in the terms outlined by Knorr Cetina (15), which especially emphasizes a hermeneutic perspective – a perspective which involves the critical recovery of the historic and social nature of any knowledge, including techno-scientific knowledge (14).

During one of our interviews, L remembered the different stages of the study of the family while re-reading Juan's medical history. That is, we were talking about the manuscript, however, L did not pay attention to the manuscript, but to the medical record which gathered the patient's medical history and its innumerable details. There he found a better base from which to explain the diagnostic process, because the medical record contains the narrative of the case, it holds the data from all the performed studies as well as the discarded or confirmed hypotheses and the decisions or actions taken regarding the patient (6).

Since he had this material at hand, one of the many emails that were sent to the US specialist came up, in which the subject of the messages was "*big hands big feet*" or "*the family big hands big feet*." This was the name used during the study of the family, before defining the disease name that appears in the manuscript.

The manuscript title defines a name for a new disease and makes its indexation possible, that is to say, it presents a final diagnosis of a *scientific nature* or that conforms to the standards of a published article in a specialized journal. However, circumstances relative to the context and the situation of the clinical geneticists can also be discovered in the name of the syndrome. Therefore, although a part of the name represents its principal symptoms, how can we explain the inclusion of the word Uruguay, the name of a nation?

McKusick (29) says that naming is the first step in defining a disease or a syndrome and that it is advisable for the name to be related to its etiology. However, the use of eponyms (proper names, geographic areas, etc.) can offer some information about the history of the disease, the contribution of the people involved in the research, its geographic distribution, and so on.

As regards the eponym "Uruguay" as part of the name of the syndrome, L and M explain:

> [...] *taking into account the difficulties we had faced and all we had experienced with this family* [...] *Because we've had experiences about, well, being from Uruguay. Like when I went to France for my scholarship and met with the professor with whom I had been corresponding by letter for over a year, he asked, "And how is Stroessner?" That is, he confused Uruguay with Paraguay. So Uruguay is this little, distant country, unknown in the world for breakthroughs in this discipline; that's why we thought it was interesting to call it the Uruguay syndrome, because having the name of a country would create in the reader the notion of at least where Uruguay is located.*
>
> *Putting Uruguay in the name also puts us on the scientific map* [...] *In Uruguay there are people capable of diagnosing a syndrome.*

In this way, the eponym Uruguay can also be considered a manifestation of circumstantial

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details related to the living and working experiences of geneticists; the bonds physicians establish with the experience of their nationality is of great interest in this regard.

CONCLUSIONS

As narrative beings (6), stories represent human beings. In fact, the products of human intellect are, to a certain extent, related to stories (30). Indeed, the starting point of our doctoral thesis, the results of which are partially presented here, is a story introduced at the beginning: the surprise – in the sense of astonishment – that we experienced when we read the Uruguay syndrome manuscript for the first time. In fact, our surprise comes partially from the strong divergence between, on the one hand, the stories and narrations referring to the different stages of research, the relationships with different family members, and so on, which we had heard about in a family context and, on the other hand, the strict and formal narrative structure of the published case report.

The knowledge of the different narrative rationalities implied was, as mentioned above, principally triggered by a critical attitude that Gadamer (24) defines as a hermeneutic philosophy interested in understanding cognitive procedures in general (14). In fact, our confusion, somehow, is resolved through acknowledging the act of construction and narrative perception required to understand the patient and reformulate the story of their disease into a medical narrative which has a place in the diagnostic taxonomy (5). In this paper we show a partial reconstruction of the study of the family suffering from the Uruguay syndrome and insist on recovering work stories based on a model of circumstantial knowledge (12) and on the acknowledgement of its importance in medical practice.

FINAL NOTES

a. The Uruguay syndrome is indexed in the database "Online Mendelian Inheritance in Man," reference code OMIM 300280.

b. A curious linguistic fact should be noted: scientific papers were first given the name of "manuscript," although today texts utilize computer processing (4). In this paper, we used the words "manuscript," "paper," "article" and "publication" as synonyms.

c. We consider that our research followed an "Ingoldian" path at its start, in reference to the English anthropologist Tim Ingold. He defines anthropology as a living philosophy of committed observation which arises as a consequence of "being" in the world in a participative conversation with its inhabitants. This idea brings to light the inevitable influence of our life experiences when choosing the methodological approximations we use to solve problems at hand.

d. Abduction is a term developed by Charles Peirce, who is considered to be one of the founders of modern semiotics. To Peirce, abduction is the process of formation of an explanatory hypothesis.

e. Clinical experience is the set of experiences obtained by a physician thanks to their ability to gather, interpret and synthesize preliminary information through observation, recognition of anomalies and their variations, which condense knowledge that is activated in the encounter with the patient (6,8).

f. However, this practical epistemology or phronesis is not explicitly acknowledged in the daily medical practice of physicians or in medical education environments (5).

g. A clue is a print, track, sign, element or signal that, because it is an involuntary result of its author, emerges as an apparently irrelevant piece of information. However, it is important in discovering a reality revealed by clues, especially for those who were educated and trained in deciphering them.

h. Chaui (13) notes that, according to Plato, Philosophy begins with admiration or, as his disciple Aristotle wrote, with wonder or astonishment: "admiration and wonder mean acknowledging our own ignorance and that is exactly why we can overcome it" (13 p.18).

i. The article was published in 2000 and to date there are no other similar cases recorded in the

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analyzed databases (PubMed, OMIM, The London Dysmorphology Database).

j. The case report published is generally written some time after the elaboration of the diagnosis and it requires some amount of processing of all the circumstantial issues related to the research in order to deliver an organized summary "ready to be assimilated and used by a reader" (6 p.101).

k. Foucault's idea of time and space (22) is recovered here because it marks the birth of modern clinical medicine and the rise of a "clinical perspective" that implies certain codes of knowledge and forms a particular experience. To Foucault, the birth of the clinical method, which enabled making pronouncements regarding an individual using a discourse with scientific structure and rational language, is bound to the development of the medical perspective in the field of signs and symptoms.

l. The physical examination may include an "instrumental" semiology, varied and complex when making use of additional tests; and a "non-instrumental semiology," which depends on the physician him or herself (2).

m. That other members of the family suffer from the same disease is fundamental for geneticists when formulating a hypothesis according to Mendelian heredity patterns likely connected to the X chromosome, in which all the analyzed cases are males related to each other through their mothers' lineage.

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