

Case Report

Beyond Diagnosis: A Case Study on Healing Time

Nadia Urli

Nursing coordinator, Pediatric Department, S.Daniele Hospital, Italy; Associate Professor, Medical Anthropology, Nursing Department, University of Udine, Italy

ABSTRACT

This study aims at reflecting about how, along one year, a Pediatric Care Team managed the case of a 16 years old girl (motor-limited since she was 15 months) who had been diagnosed a rare disease (that is, Ehler Danlos Syndrome). Such experience inspires observations about the meaning of diagnosis, the organic or psychiatric reading of symptoms, the effectiveness of the therapy and the perception of competence tested out by the Caring Team. A multi-professional comparison proved necessary, yet both assessment and treatment were left to the Pediatric Team – which required a deep analysis of the care strategies. We witnessed real suffering, yet we realized that the case history did not correspond to the expected evolution: though the diagnosis was never under discussion, we preferred working within the caring space – so that the girl

was accompanied along a demanding path of medical advice in which symptoms evolved from pain to lipothymy to scattered tremors. The team agreed on the relevance of a time for care. If we had limited our reading to the symptoms, the girl would have entered the vicious circle of either psychiatric care or chronicity. On the contrary we proposed her a non-medicalized context that included group advice and educated the family to a ‘normalizing’ management of their daughter. As a result, the girl resumed her everyday life – which taught her that problems can be faced, instead of being simply suffered.

MeSh Headings/ Keywords: Rare diseases; Therapeutic effectiveness; Lived body; Symptoms language; Caring strategies

Introduction

This case proved very demanding for the Pediatric Care Unit, so for both doctors and nurses. No matter how serious the situation, or whether its solution apparently limits to lowering the inevitable impact of a chronic disease, it showed that focusing on the patient (rather than on the disease) gave the former unexpectedly positive results; it proved the latter is always possible to heal [Appendix 1]; and it taught both doctors and nurses how to elaborate a taking charge strategy for disabled patients with high medical needs.

Disease is a human experience lived through one’s body. Medical anthropology distinguishes the ‘physical body’ (as described by scientific texts) from the ‘lived body’ (as known by patients and inhabited by illnesses): the latter is actually tamed by pain and meant to become a dominion for medical practice, in the name of a therapy able to ease its suffering [1]. In taking care of a patient, also nursing sciences acknowledge that the complexity of the lived body cannot be reduced to biology only: as a matter of fact, the body of a suffering person is always the means to live in the world – so it can never be reduced to a physical item or quantified as a pathological symptom [2]. From this point of view, pain is not only a symptom but also a human experience that accompanies illness: when particularly acute, pain turns the person helpless and vulnerable – so into a condition one is never prepared for. Before acute and recurrent pain, the person as a whole is in pain and his/her life project proves endangered [3].

Within this paradigm, the medicalization of the ill body presumes to confine the disease in a single anatomical district – which hinders the patient from fighting the ‘overall destruction’ that overwhelms his/her person [1]. The following story

describes the illness experience that imposed a two years’ stop in the life of a young woman (hereinafter referred to as Diana) as well as the healing plan shared with her family in order to give her back that chance of a normal life the disease had endangered.

The Disease

Ehlers-Danlos Syndrome (hereinafter, EDS) is a group of relatively rare, genetically heterogeneous connective tissue disorders caused by a quantitative deficiency of fibrillar collagen. Affected patients appear normal at birth – only to develop skin hyperextensibility, joint hypermobility, and/or tissue fragility [4]. EDS has been classified into 6 clinical forms. The classical form is characterized by joint hypermobility, scoliosis, mitral valve prolapse, easy bruising, skin hyperelasticity and fragility: this disorder has an autosomal dominant inheritance [5]. Hypermobility EDS shows through generalized severe joint hypermobility and minimal skin manifestations [6]. Vascular EDS is characterized by pronounced dermal thinning and risk of cardiovascular accidents [7]. Kyphoscoliosis EDS is manifested through joint hyperextensibility, hypotonia, kyphoscoliosis, fragile cornea and bones. Arthrochalasia and dermatosparaxis are more rare forms of EDS. Psychological distress is a known feature of generalized joint hypermobility as well as of its most common syndromic presentation (that is, hypermobility type) and it significantly influences on the quality of life of affected individuals [8].

Meeting Diana

Diana is 17 years old and attends a private, quite prestigious high school: she has always been deeply involved with sports and cultural activities, and she has always been brilliant

in her studies. Yet, since she was 10, she has also got a long and complex story of syncopal fits, migraine and limb acute pain and hypotonia. Along the year prior to the meeting, pain symptomatology increased: this strongly limited Diana in her relationships and schoolwork – though the latter was carried on quite positively. Her family was very careful and accompanied her along the medical tests that ended with the EDS diagnosis – one year before her first admission: since Diana had been using crutches for months and had suffered several pain fits that brought her to the First Aid (prior being admitted to Hospital), a physiotherapeutic cycle was suggested along with a painkilling therapy (including opiates). Her doctor requested her admission in the early autumn 2014 in order to achieve clinical stabilization: she reached the ward on a stretcher as she could not walk. Admissions spanned one year: on the whole, 6 months spaced out by three discharges.

A Paralyzing Pain

Taking charge of Diana had a double aim: easing acute pain and scheduling a basic pain treatment. The first admission lasted one month and a half. The early days passed silent in a penumbral room, between short moments of well-being and her parents overcome with tangible anguish: her pain was so absolute as to prevent any everyday activity and nailed her in bed. Diana's days were marked by intravenous painkillers, whose effect used to fade far too early – whereas her parents started to put questions about the efficacy of a clinical strategy that seemed to lead nowhere. This complain of theirs counterbalanced the impotence experienced by the healing team itself: what else could be done, since the specialists in rare diseases had not been able to achieve any stabilization? Which therapy could be devised for this young woman who kept on planning her life (from present education to future occupation) in spite of a diagnosis that had apparently shattered her biography? So constant was the use of opiates to arise an ethical dilemma about whether such therapy really was in Diana's behalf: unfortunately she was always sleepy, which paralyzed the therapeutical action – whereas her disease was so peculiar as to instill doubts about how to proceed.

The healing team thus consulted the anesthetists as specialists in the pain therapy: the aim was to limit the use of morphine through a local treatment of symptoms (so, by injecting bupivacaine and dexametasonone into the myofacial triggers). Parallel to the professional advice from the ESD specialists, just one strategy was suggested: pain control and physiotherapy, though its start was repeatedly postponed due to a deafening pain Diana tried to hush by moving her body. The healing team also wondered why such a young woman was hit by this disease, knowingly invalidating in the long run – and why she went through and prematurely experienced its most critical phases. Such a feeling continued puzzling the team till the day they decided to postpone an umpteenth request of opiates (much exceeding the admitted dosage), only to turn to the symbolic power of the pharmacological therapy [Appendix-2]: in other words, to use a physiological solution – a neutral substance with no therapeutic effect. Diana's pain disappeared and a bridge was

built: on the one hand, this led Diana to a new reading of her own pain; on the other hand, this required ethical responsibility in handling such discovery – that is, in informing her parents and asking for their help to better understand the situation. Yet her parents too had incorporated the diagnosis (and, through it, that rear and invalidating disease): as a consequence, getting their help depended on helping them through a new comprehension of reality.

The healing team thus started a series of meetings with professionals, with Diana and with her parents – in order to contribute acceptance of the disease symptoms. The team themselves started a supervision schedule with a colleague in psychiatry: he was asked to help Diana by 'normalizing' her life, increasing her pain threshold also through low dosage neuroleptics (such as olanzapine and paroxetine) and, above all, learning to 'read' the messages from pain. The new therapy now included phases of active listening and participant observation meant to detect the elements inducing pain, whereas the touch was chosen to ease muscular tensions: as for the symbolic meaning of both therapy and its giving, it was used to guide her to oral treatment only. The benefit was immediate: Diana passed from morphine to painkillers (paracetamol), her friends kept visiting her and also her appetite bettered – which slightly increased her weight weight and permitted the start of physiotherapy. As she was discharged, she was planned a return to her usual activities as well as advised not to resort to the emergency service in case of episodes with minor impact and no complication: in alternative, she was suggested to contact the ward by phone. The healing team kept waiting for the situation to develop, as relapses were likely to take place: on the one hand, both the family and the team shared the caring strategy; on the other hand, Diana still lingered on in incorporating an invalidating disease – which made her way back to life quite demanding.

The Evolving of Symptoms

Diana's home stay lasted about four months: she resorted to the doctor's surgery for more and more frequent and lasting faints, but she also led quite an average life – thus attending school, seeing her friends and riding her bicycle. She was rushed to hospital when faints caused losing consciousness, falling to the ground, nausea, tremors and uncontrollable leg pain. Diana's days so swung between moments of well-being (when her good mood matched with appetite and walks) and moments of deep crisis (when loss of consciousness, sweat and paleness matched with a new symptom – that is, her body kept shaking). Such episodes did not alter her vital parameters: it all apparently expressed on the outside (from the skin to the body movements), yet the end of such fits let Diana catatonic.

This proved a most puzzling moment for the professional team too: had there been anything wrong with the therapeutic approach? Did the new project give Diana any benefit? Was it necessary to turn her to a specialized center? A new consultation with the specialists confirmed the strategy: but it also proved there was nowhere to send such patients. The healing team could only integrate its medical analysis with two diagnostic

tests, which generated an ultimate dilemma: if the therapeutic plan offered no benefit, did it make any sense to resume any diagnostic strategy? Was it ethical to propose them to both Diana and her family, thus raising empty expectations? Once again, a healing action showed a possible way out: during one of her fits, a delicate foot massage was practiced, in order to reassure her and make her feel she was not alone. As a result, the body region did not shake when massaged – which made it possible to repeat such result for short fits, while waiting for their spontaneous end. In spite of the development of Diana's syndrome, the healing team carried on this strategy – due to their firm belief everything was to be done in order to grant her as many years as possible. A colleague in psychiatry helped the team in the relationship with her family: the aim was to depathologise so 'eloquent' symptoms.

Across the Line

When Diana's fits kept lasting up to one hour, and as the doubts about the efficacy of the treatment strategy re-emerged, the professional team chose to complete their diagnostic tests. Diana was moved to the Intensive Care Unit where she could be constantly monitored: an EEG analysis also excluded the occurrence of any epileptic fit. While in the ICU, Diana was daily visited by the paediatric team: she looked pale and was usually sedated; when conscious, she looked confused – a young woman and an alien among the other patients, all of them adult and seriously damaged. As losing her was a major risk, she was moved to the Care Unit as soon as possible – and there Diana experienced repeated, though shorter fits – so the healing team activated a strategy of psychological containment [Appendix-3]: during the fits, the girl was spoken to because she was able to hear the words; when the fits were over, she answered precise questions and never looked upset – yet she proved rather silent than ready to share her latest experience. When offered a psychologist, she refused him and turned less receptive to the professionals who had introduced him.

The healing team then wondered about the meaning of such refusal Diana seemed to credit with a specific meaning, but they also felt the ethical responsibility of trying any approach: so they joined the psychiatrist in evaluating the opportunity of an alternative strategy that could keep them in touch with her. A relationship meant for help cannot include any 'like-vs-dislike' mechanism or any skipping of the situations the team feel the patient's hostility: hence, the healing team continued with their intervention without questioning her past – which corresponds to the professionals' principle of responsibility. They chose a strategy of positive examples based on real-life narratives whose message was 'a problem can be solved': if healing also includes dialogue, it was high time to use words not only to support or quiet Diana, but also to lead her across a line she had just discovered. The professional who volunteered belonged to the same group who had offered her a psychologist: but he also knew a fitting life narrative, so he asked Diana for her permission to proceed. The story was real: it told about a girl who learned how to take full control of her life and was told after the narrative medicine method. Diana followed the whole narration with the utmost attention and appreciated such

approach: a few days later she even started to ask when she could be discharged, as she had planned a seaside holiday with her friends. The supporting meetings continued after her ultimate discharging, in order to help her in turning back to her studies and everyday activities. Two months later, Diana was able to walk again without any aid and to do nearly everything she had to hold on due to her illness. A few, short fits still occurred, but she had grown able to cope with them with the most functional strategies.

Case Analysis

The territorial Pediatric Unit that took care of Diana focusses on primary treatments: dealing with a "rare disease" on a specialized level so implied dealing with treatment models based on other therapeutic approaches. Diana's story thus helped to detect two thematic levels: on the one hand, the medical method itself was examined as for the meaning of diagnosis, the encoding of symptoms and the use of clinical procedures; on the other hand, the relation between general medicine and specialistic medicine (all over its several levels) was analyzed. Far from being an exact evaluation, objectifying a pathological state (through both diagnosis and the synthetic and approximate description of the patient) proves rather hard. The criteria on which such diagnoses are based prove useful on the practical level, but they cannot expect to wipe out the differences among the patients involved. As in Diana's case, the descriptions labeled as 'syndromes' are rather clear: which suggests the use of dynamic diagnosis – that is, the same approach when dealing with mental health [9]. Whereas this is the present state of the medical art, a rare disease diagnosis somehow puzzles the healing practices: in other words, when trying to control the course of the disease (in particular if its signs have a strong emotional impact), the search for a leading rule proves problematic [10]. This affects the encoding of the visible signs of a disease: in describing both symptoms (so, subjective disorders) and signs (that is, the changes objectively noticeable by a doctor), semiotics has joined the progress in medical technology to bring knowledge into depth – but it still requires the ability to interpret and integrate it into a more complex diagnostic synthesis [11]. Modern medicine has undoubtedly improved as for 'doing', yet this should not come to the detriment of 'thinking' – since patients could feel lost in reflection [12].

Starting from the patient's suffering as a whole, the healing team is asked to detect the main organic problem and to further study it through step-by-step break-up: a most typical method of specialized medicine, in particular on the third level where rare disease studies belong [13]. Nonetheless, the issue remains about how to combine wide and deep competences with a clinic competency bringing together various levels of know-how: that is, about how to set the patient within a frame more respectful of his/her illness [14]. The medical literature, consulted about the Joint Hypermobility Syndrome (EDS), stresses the mind-body interaction [8] and acknowledges the link between the disease and a range of stress signs (such as chronic fatigue syndrome, chronic pain, irritable bowel, pressure alternans and vagal symptoms) and psychiatric disorders (from anxiety and mood disorders, to autism). As a matter of fact, and in spite of such a wide range of possibilities,

Diana had been offered a therapy based on symptoms only: that is, easing of acute pain and scheduling a base anti-pain treatment. Yet doubts remained about such approach: would this suffice to have her walk out of the hospital? Could she get back to her life after joining a psychiatric support project, or would a non-psychiatry-based context be more profitable? On the whole, Diana's case exposed a mixture of uncertainty and responsibility in making therapeutic decisions: above all, for a 'first level' specialized team – who had to confront with both the perception of their own competency and the ethicality behind the treatments they suggested.

Conclusions

As shown by Diana's story, dealing with such an amount of meanings (conveyed through the lived body) urged the healing team to reconsider their convictions about therapy as a symbol: in this case, they turned to a placebo as a bridge to relate with Diana and her family, who used to feel 'prisoners' of both pain and their useless attempts to control it. Once any organic cause was excluded, it would have been easy to label the symptomatic cluster as psychiatric – yet such a problematic quest for meaning imposed a change in communicative register: symptoms needed analyzed as a significant whole, whereas the individual (rather than the disease) needed studied. Once the known healing strategies failed, also the field search in caring required a substantial investment in energies: this included participant observation; a repeated daily confrontation, aimed at integrating Diana's microscopic behavior; the personal freedom to use one's own positive talents; and the prevention of a bent to expel as a temptation set by difficulty. Such delicate monitoring of the relationships within the healing team proved useful to tell one's feeling of impotency and to transform potential implosion into an opening to conversation with the other healing operators.

Group counseling made it possible to take advantage of alternative therapeutic strategies that were not typical of the pediatric context (such as the non-pharmacological containment, the decoding of implicit communication and the giving of drugs through its symbolic value): all of these strategies were available at the Mental Health Department, so they were stored by the healing team and used to contain the anxiety about their own initial perception of lacking competency. This way, Diana and her family could benefit of all the trustfulness, time and quality presence they needed. Such place for reflection hosted repeated weekly conversations, where operative difficulties could be discussed and Diana's behavior possibly encoded. All over these passages, the healing team always kept the control over Diana's case: which proved most strategic in such a complex situation, since separate consultations might have generated a fragmentation of both the healing project and the life of a young woman who had already embodied too many fragments.

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ADDRESS FOR CORRESPONDENCE:

Nadia Urli, Nursing coordinator, Pediatric Department, S. Daniele Hospital, Italy; Associate Professor, Medical Anthropology, Nursing Department, University of Udine, Italy, E-mail: nadia.urli@gmail.com

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Appendix

1. The Italian word *cura* (translates both the English ‘care’ and ‘cure’ so it lives on nuances that need explaining. The scientific literature about nursing distinguishes ‘cure’ from ‘care’: the former corresponds to ‘curing’ as ‘treatment’; the latter means ‘taking care of’, hence ‘caring’ as a basic notion of the nursing sciences – in that it interacts with the concepts of ‘empathy’ and ‘therapeutic relationship’, thus being the core of professional nursing [15]. As for ‘healing’, anthropological literature endows it with a wide acceptance: as a set of gestures and words, healing is performed through the support, help and protection of people weakened in body and soul by permanent or momentary limitations depriving their social lifestyle of ‘normality’ or ‘independence’ [16]. Hereinafter, the Italian word *cura* is meant as ‘healing’.
2. Giving a drug means offering a pharmacological substance that produces a therapeutic effect: nonetheless, in this action medical anthropology also detects a symbolic message through which the patients understand s/he is being taken care of. Especially when pain and anxiety are at play, patients often (though implicitly) ask for help while (explicitly) asking for a drug: no surprise a successful counseling has an intrinsic therapeutic effect and reduces the need for drugs – especially when the patient is dominated by anxiety. This notion inspires the therapeutic meeting also in non-psychiatric contexts – and it is mainly led by non-medical operators.
3. Psychological containment implies the use of the communicative potential of counseling: the latter is recommended to the sanitary staff as a substitution of pharmacologic containment.