## Chapter 20. Stretched beyond the limit: well-being and functioning in patients with Ehlers-Danlos syndrome and other hypermobility syndromes

R Geenen and MA Lumley

Rinie Geenen, PhD, psychologist
Professor and head of the Psycho-rheumatology research unit
Department of Clinical and Health Psychology
Utrecht University
Department of Rheumatology and Clinical Immunology
University Medical Center Utrecht
PO Box 85500
3508 GA Utrecht, The Netherlands
Telephone +31302534916
E-mail R.Geenen@uu.nl

Mark A. Lumley
Professor and director of clinical psychology training
Department of Psychology
Wayne State University
Detroit, Mi, USA
Telephone 313-577-2247
E-mail mlumley@wayne.edu

## 1. Case: Michelle

Michelle is 34 years of age. She was raised in a farming family and had a generally positive and supportive childhood. At the age of 18, she moved to the city to attend university. For the past ten years, she has been happily married and has 2 young daughters. She has long realized that she has very flexible joints, and she regularly dislocated her fingers, but she was not that concerned and did not consider it to be a disorder. Two years ago, however, her situation worsened when she dislocated her shoulder while playing tennis, and the subsequent operation left her with various complications. The surgeon referred her to other specialists, and while still in the hospital, Michelle was diagnosed with Ehlers-Danlos syndrome (EDS). She was mostly pleased to learn of this diagnosis, because it explained her frail joints as well as the pain and fatigue that had been interfering with her functioning at home and work during the past few years. These symptoms had been invisible, but now others could understand why she had not been feeling well or functioning optimally. Michelle also was thankful that the hospital physiotherapist helped her learn to use her joints in the appropriate way and manage dislocations.

Lately, however, Michelle has become somewhat depressed and worried. She has come to realize that having this disease could greatly change her life. Although she has many abilities and loves her work, she fears that she will not be able to fulfil her high ambitions. She had always been able to hike in the mountains while on holiday, but this has become more difficult, and in the near future, she expects that she will no longer be able to walk even the easy trails. She needs to change so many things and is anxious that her condition will become dramatically worse in the future. She is also worried that her children have EDS and she needs to remind herself to avoid raising them too cautiously or fearfully. Although she expects that her optimistic nature will finally help her to deal with the consequences of EDS, a psychologist might help her with her current negative mood, and she is considering seeking cognitive-behavioural therapy.

## 2. Introduction

Connective tissues maintain normal bodily integrity, but when connective tissues fail in this role, multiple problems occur. EDS is a heterogeneous group of heritable connective tissue disorders characterized by articular hypermobility, skin extensibility, and tissue fragility.[1] This disorder is chronic, varies in severity from mild to very serious, and can be lethal. Hypermobility syndromes featuring joints that easily move beyond the normal range expected for a particular joint, are a threat to well-being and functioning. What is said about EDS in this chapter mostly applies to hypermobility syndromes as well.

Recently, the two conditions benign joint hypermobility syndrome (BJHS) and EDS hypermobility type have been recognized as one and the same clinical spectrum ranging from apparently symptomatic generalized joint hypermobility to the most disabled individuals fitting the new diagnostic criteria. These new criteria are more strict than the Villefranche criteria and the Brighton criteria for BJHS in order to define a homogeneous phenotype for management and scientific purposes. Within the new EDS nosology, its name is hypermobile EDS (see chapters 2 and 5).

### 2.1 Burden of EDS

The psychosocial burden of EDS is substantial[2-4], and can be summarized by the central theme of "living a restricted life".[5]

First, EDS is a relatively rare condition, which means that the diagnostic process is often protracted and confusing, and that healthcare providers often have insufficient knowledge of the medical problems and the possibilities for treatment and care.

Second, people with EDS have fewer opportunities in daily life than healthy people. For example, pain, fatigue and other symptoms hamper functioning, and people with EDS may fear articular dislocations or skin damage, which further limits activities.
Third, patients may experience invalidation, or stigmatization and lack of understanding from family and friends, health care providers, co-workers and others in daily life.
Finally, given the heritable nature of EDS, the pros and cons of parenthood are often evaluated, and decisions to refrain from childbearing can be highly disappointing.
Psychological evaluations and interventions for patients as well as their families are vital to help them cope effectively with this disorder.[6] Self-care, counselling and therapy are directed at both the prevention and treatment of adverse consequences. Because most bodily organs have connective tissue potentially affected by EDS, multiple bodily system problems can occur, and virtually all medical and allied disciplines play a role in education and treatment.

### 2.2 Aim

The aim of this chapter is to review the somatic and psychosocial consequences of EDS; that is, the effects of EDS on physical, psychological, and social well-being and functioning. Somatic problems, psychological distress, and social functioning are reviewed, and suggestions for coping with these problems in everyday life are given. We base our discussion and recommendations on the relatively few empirical articles on psychosocial functioning in EDS, supplemented by our knowledge of experiences of individual patients, and our knowledge of the somatic, emotional, and social consequences of chronic disease more generally.

## 3. Somatic problems

In a Swedish survey using the Subjective Health Complaint Inventory (SHCI), 99% of 250 participants with EDS reported health complaints with a mean number of sixteen complaints for the whole sample.[7] The somatic problems reported most often were musculoskeletal by 246 (98%), pseudoneurological by 241 (96%), gastrointestinal by 236 (94%), allergic by 182 (73%) and influenza-like by 144 (58%) persons. These specific somatic problems as well as those of vascular EDS are discussed in other chapters of the book. In the current chapter, pain, fatigue, joint dislocations, fragile skin, and sleeping problems are discussed.

### 3.1 Pain

In a questionnaire survey of 41 patients with EDS, 63% of the patients reported relatively elevated pain (above the scale mid-point), and only two patients reported having no pain.[8] Another study used structured interviews of 51 patients with EDS and found that 90% experienced pain for at least six months.[9] In a larger questionnaire study among 250 patients (contacted through the Dutch patient association for EDS), pain during the past week was reported on a 0 to 100 mm visual analogue scale (VAS), 100 being the maximal level of pain: 29% of the patients reported pain between 0 and 25, 24 % between 25 and 50, 28% between 50 and 75, and 18% between 75 and 100.[2] Of this sample, 41% reported chronic, widespread pain; that is, pain existing for more than three months in the upper and lower, left and right, and axial parts (i.e. spine) of the body. Of the respondents, 55% commonly used analgesics—mostly paracetamol (acetaminophen), non-steroidal anti-inflammatory agents, and tramadol. One study found that pain is more prevalent and severe in hypermobile EDS than in classical EDS, and that pain is associated with hypermobility, joint dislocations, previous surgery, poor sleeping quality, and functional impairment in daily life.[10]
Thus, pain clearly is a prevalent problem in EDS, and has consequences for well-being and functioning. Reduced well-being and functioning, in turn, may contribute to the persistence and severity of pain. Education and self-management techniques likely can help to address

relatively low levels of pain, but more serious pain should be treated in cognitive-behavioural therapy (for explanation, see Box 20-1) and physical rehabilitation programmes. Such therapy is especially indicated if pharmacological treatment is ineffective to deal with pain and its consequences.

**Box 20-1** Cognitive-behavioural therapy
A main premise of cognitive-behavioural therapy is that negative, dysfunctional thoughts have a perpetuating role in health problems, and that people need to learn skills or techniques to change their thoughts and feelings. Cognitive-behavioural therapy is directed at reduction of symptoms like depression, anxiety, pain, and physiological responses by changing maladaptive thoughts and actions. Examples are interventions with one specific aim—for example, relaxation, stress reduction or overcoming of fear-avoidance beliefs to support an exercise intervention and, more commonly, the incorporation of various methods—for example, cognitive restructuring of dysfunctional beliefs or "worry" thoughts, pain coping skills training, activity pacing, stress management training, relaxation exercises, exposure to anxious situations, thoughts and worries, and positive self-talk.

### 3.2 Fatigue

The survey of Dutch patients with EDS asked respondents to note the five most serious symptoms or problems of EDS.[2] Fatigue (64%), pain (54%), skin fragility (33%), and problems with the back (29%), wrist, hand, and fingers (27%) were mentioned most frequently. It is striking that so many patients reported fatigue among their five most serious problems. This survey also compared general fatigue scores in 250 patients with EDS to those of healthy people from the general population using the Multidimensional Fatigue Index.[11] The occurrence of 'general fatigue' above a specified cut-off point was five times larger for patients with EDS than for people in the general population.[2] A similar survey of 274 Dutch patients with EDS, which was conducted five years later in the same population, found that 77% of the patients reported severe fatigue as compared to 11% and 17% in two predominantly female samples of the Dutch general population.[12]

One possible way to address the elevated fatigue experienced by so many patients with EDS is to identify specific patient and disease characteristics that contribute to fatigue, and to change targeted lifestyle behaviours. For example, it has been proposed that aerobic fitness or strength training to deal with muscle weakness, or using weight management techniques to reduce obesity to treat sleeping apnoea, might be useful approaches to reduce fatigue in joint hypermobility syndromes.[13] Unfortunately, the physiological origins of chronic fatigue are unknown, and there are no effective pharmacological treatments. Thus, although fatigue is indisputably an adverse consequence of EDS, it is most sensible to consider behavioural means, such as life-style adjustment and cognitive-behavioural, physical exercise, and sleep hygiene interventions.[14]

However, the effects of such behavioural interventions have not been investigated in EDS. Caution is needed; for example, physical exercise programmes need to take account of possible joint dislocations as potential complicating factors. Moreover, to enhance their sense of vitality, patients will need to find a balance between physical effort and rest. Some patients may avoid all physical exercise to prevent pain and fatigue, but this will promote physical deconditioning. Other patients may persist in exercising despite pain and fatigue and stop only when pain or fatigue are extreme. However, this may over time result in dramatically reduced physical activity, as stopping exercise is stimulated because it takes away pain. Often a gradual build-up of the intensity of physical activity is important to prevent relapse. As in other chronic symptomatic conditions, activity-rest cycling can be valuable, with patients finding the optimal timing of activity and rest. A predetermined activity duration—rather than

pain or fatigue—should determine the timing of the rest-activity cycle. More generally, some patients will need encouragement to exercise, whereas others will need to learn to stop exercising in time.

### 3.3 Joint dislocations

In some forms of EDS, from 90 to 100% of the patients, report hypermobility of the joints[15], but the severity of joint problems differs considerably among individuals and EDS types. Joint problems hamper work, sports, and many other important daily activities. Patients need to find an optimal balance between living passively to avoid joint dislocations and a more active - but risky - lifestyle, which can trigger joint dislocations. The challenge for patients is to learn to use their body without pushing their joints beyond their limits. Most patients learn this through trial and error. It is important that patients do not avoid physical activity, but instead use assistive technology, adaptations at home and at work, and aids such as braces and appropriate shoes to remain physically active. Fellow patients, rehabilitation physicians, and occupational therapists can help the patient to find adjustments that help in optimal functioning.

### 3.4 Fragile skin

Fragile skin is a problem in 40% (hypermobility EDS) to 100% (classical EDS) of the patients with EDS.[15] Wound healing may be problematic, and wide, ugly scars may result. Our society values beautiful skin, and physical appearance is important not only for self-esteem but also for interpersonal communication, especially first contacts. Patients can try to prevent skin damage by wearing appropriate clothes and by avoiding risky situations. In case of wounds, appropriate care is important. In serious cases, wound care nurses, dermatologists, or plastic and reconstructive surgeons can be helpful.

### 3.5 Sleeping problems

The prevalence of sleeping problems in EDS has rarely been studied. In a small sample of nine patients with EDS, five reported sleeping problems, and six reported periodic limb movements.[16] In contrast to this seemingly high prevalence, the Dutch survey of 250 patients found that only 4% reported sleeping problems as among their top five most common problems, but this report is likely biased by the method of requiring patients to pick a maximum of five major problems.[2]

Potential sleeping problems should be assessed in patients, and if present, differential diagnosis of the aetiology of the problem should be pursued, because treatment for sleeping problems should preferably be aimed at the cause. In the differential diagnosis, sleeping apnoea has to be taken into consideration, because as soft tissues weaken by becoming older, especially in EDS patients sleeping apnoea can become an extra problem.

Sleeping medication is generally not recommended for patients with EDS because of the resulting muscle relaxation and the ensuing increased risk of joint dislocations. Insomnia can be treated with stimulus control therapy, relaxation training, and cognitive-behavioural therapy.[17] Such interventions may include techniques such as avoiding the consumption of coffee, alcohol, and heavy meals within several hours before sleep; making the bedroom dark, comfortable, and quiet; using the bed only to sleep (and for sex), thus not for reading and watching TV; having a regular pattern of effort and rest throughout the day as well as a fixed sleeping time; and - if needed - using stress-reduction, relaxation or thought-stopping techniques. Insomnia can also be treated with sleep restriction therapy, which reduces the patient's time in bed to the total number of hours of sleep that is being obtained, and forces patients to be up and out of bed when not sleeping; as sleep efficiency increases, the sleep period can be lengthened.

## 4. Psychological well-being

Psychological well-being is a core indicator of quality of life. The severity of one's disease influences psychological well-being, but it is only one factor, and the relationship between disease severity and well-being is weak in most chronic diseases.[18] Coping, resilience, emotional and social factors play larger roles in adaptation and well-being.

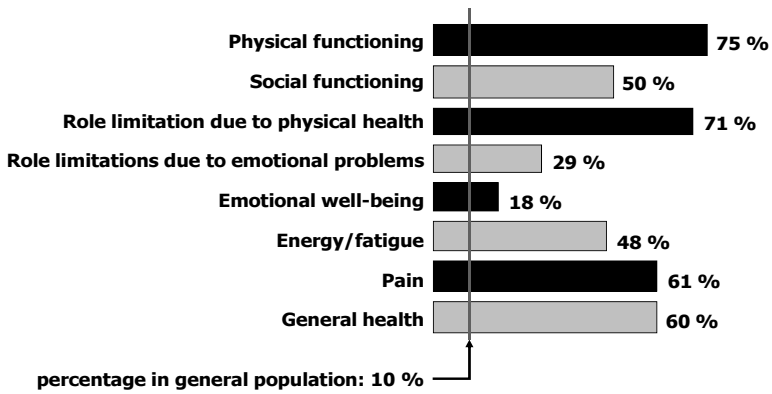
### 4.1 Emotional consequences

Negative emotional consequences of living with EDS are common. Interviews show that some people with EDS live with constant fear of skin damage, physical deterioration because of hypermobility, unemployment, and problematic pregnancies.[5] Patients with joint hypermobility syndrome present with greater prevalence of panic disorder and agoraphobia.[19,20] Parents who have children with EDS, live with the fear that authorities, medical personnel, or others will suspect them of bad parenting or abuse when their child presents with a joint dislocation or bleeding[21], and even if no accusation are made, parents may still sense suspicion.[5] Another problem mentioned in interviews is the reduced possibility of developing one's abilities.[5] The great effort needed to achieve something substantial in education or work is often not possible in case of EDS. This especially holds for people with vascular EDS who often know at a young age that, although new medication has improved the prognosis, their life expectancy is shortened, which has implications for important life choices in education, work and family planning; and can impede life satisfaction.

A questionnaire study of 41 patients with EDS from North America found that anxiety, depression, anger and relationship worries were quite high.[8] Indeed, these problems were seen three to four times more frequent than among the general population. Interviews of these patients revealed that patients attributed their emotional problems to chronic pain, impaired task performance, reduced social contacts, sexual problems, worries about having children, and frustration with the medical system. Of the 41 patients, 56% had had psychotherapy, and 46% (had) used antidepressants or anxiolytics. These estimates of emotional difficulties in EDS may be relatively high, probably because this sample was recruited for a special research study that likely attracted very motivated or distressed patients from across the continent.

The Dutch survey showed less emotional disturbances among people with EDS than did the North American study. In analyses using the RAND SF-36 questionnaire, the cut-off defining unhealthy functioning was set at the 10th percentile of functioning of the general population.[2] According to this admittedly arbitrary criterion, 10% of the general population shows poor functioning on a dimension of quality of life. Among patients with EDS, the percentage of poor functioning on emotional well-being was 18%, whereas 30% reported experiencing role problems secondary to emotional disturbances (figure 20-1). Overall, the risk of poor emotional well-being was twice as high as normal.

**Figure 20-1** The percentage of 250 patients with Ehlers-Danlos syndrome having a low score on eight aspects of quality of life *




** The criterion for a low score was set at the 10th percentile of the general population. According to this criterion, 10% of the Dutch population obtains a low score on the RAND SF-36 questionnaire.[46]*

### 4.1.1 Depression

This Dutch survey also included the Zung Self-rating Depression Scale.[22] The scores were compared with scores of a healthy control group (although this is not reflective of the general population, because patients with a chronic disease were purposely excluded). As compared to this healthy group, 48% of the patients with EDS were classified as having a depressive mood if the 10th percentile of the control group was taken as cut-off criterion.

The approach to treating depression should depend on its severity. Antidepressant medications and more extensive psychological therapies, such as long-term cognitive-behavioural therapy, are recommended to treat depression after simpler methods (e.g., guided self-help or exercise) have failed to produce adequate responses. Combining pharmacotherapy with evidence-based psychotherapy typically provides a modest increment over either treatment alone.[23,24] In both severe and mild depression, mental health specialists, particularly clinical psychologists and psychotherapists, should provide cognitive-behavioural or interpersonal psychotherapy to prevent relapse or recurrence.[23,25,26]

### 4.1.2 Anxiety

For patients with elevated anxiety, selective serotonin reuptake inhibitors (SSRIs) generally are the most common pharmacological treatment.[27] Of the non-pharmacological therapies available, cognitive–behavioural therapy is the preferred first-line treatment for anxiety disorders.[28] This approach employs a variety of techniques including cognitive restructuring, exposure to anxiety-generating stimuli, and behavioural experiments.[29] The rationale behind cognitive restructuring is that intense, persistent negative emotions (including anxiety) and maladaptive coping behaviours (e.g. avoidance or safety-seeking behaviours) follow from how these situations and events are appraised or perceived. Accordingly, to alleviate emotional suffering and foster constructive coping, it is important to: (a) identify the maladaptive cognitions that underlie a person's anxiety in particular situations and

circumstances; (b) examine the validity and utility of these cognitions; and (c) change these cognitions into those that are less anxiety-generating and that facilitate rather than block constructive action. In the treatment of panic symptoms, for example, cognitive restructuring is often targeted at altering catastrophic misinterpretations of benign bodily symptoms, which the patient otherwise appraises as dangerous. Likewise, in a person with social anxiety, restructuring could focus on excessively negative inferences about how one appears to others.[30]

### 4.2 Coping

Chronic illnesses such as EDS are often accompanied by a helpless or passive stance by patients, who feel unable to influence or master their well-being. Helplessness is often expressed in global negative beliefs such as "because of my illness, I miss the things I like to do most," "my illness controls my life," and "my illness prevents me from doing what I really like to do".[31] These thoughts are often automatic in response to one's illness and negative events. Such helplessness and avoidance of attempts to influence outcomes not only contribute to suffering but also increase the risk of anxiety and depression.

In contrast, effective coping affects the impact that EDS has on psychological well-being. Some research of coping and related constructs with EDS has been conducted. Psychological well-being is increased in people who demonstrate either acceptance or a sense of coherence.[32] The latter refers to having confidence that bodily reactions as well as environmental stressors are predictable, and that one can handle the negative consequences of these stimuli. In general, low helplessness and high acceptance reflect that a patient is able to deal with the consequences of a chronic disease.[31]

For helplessness and negative automatic thoughts, cognitive-behavioural therapy (Box 20-1) can help in recognizing, discussing, and restructuring of these thoughts, setting of goals, and practicing steps to reach these goals. The dual-process coping model stresses the fit between characteristics of the situation and the specific coping strategy used.[33] The restructuring of cognitions and behaviour can help to deal with situations that can be changed, whereas acceptance of the inevitable consequences of the disease needs to be part of patients' coping repertoire to deal with situations that cannot be changed. The ideal response of a patient is to use both assimilative ways of coping (i.e. active attempts to alter an unsatisfactory situation in a way that fits personal goals and aspirations) and accommodative ways of coping (i.e. the adjustment of personal goals and aspirations to current situational limitations in order to accept the situation or interpret the given situation less negatively). Such an approach will help the patient to change life circumstances as needed while maintaining a satisfying life perspective.

In recent years, mindfulness-based stress reduction therapy and acceptance-based therapies have been applied in the treatment of mental problems that may accompany chronic diseases.[34] These therapies add mindfulness and acceptance to traditional cognitive-behavioural techniques. Mindfulness meditation focuses on becoming aware and accepting all thoughts, feelings, and sensations instead of trying to avoid or fight them. Reviews have found statistically significant, small to moderate effects of mindfulness-based therapies on psychological distress, pain, and coping behaviour.[35-37] Integrated mindfulness and cognitive-behavioural therapy may enhance the treatment efficacy.[35]

Traditionally, psychologists and other mental health providers have focused on patients who were unable to adequately adjust to the consequences of chronic diseases. In recent years, the emphasis on the reasons why people fail to achieve a healthy adjustment has shifted to the identification of factors that help patients make that adjustment. To promote psychological adjustment, patients with chronic diseases are encouraged to remain active, to acknowledge and express their emotions in a way that allows them to take control of their lives, to engage

in self-management, and to focus on potential positive outcomes of their illness.[38] Patients who use these strategies have the best chance of successfully adjusting to the challenges posed by a chronic illness like EDS.

## 5. Social functioning

Support from friends and family is potentially an important influence on how well people manage and cope with their disease. In research, this response of others has been termed "social support"[39], and it has been conceptualized both structurally—that is, the size and composition of a person's social network, and functionally. Functional support consists of actually "received" support (also called enacted support), which refers to what the others actually do or provide for the patient during times of need, and the "perceived" social support. It is the perceived support, which appears to be most closely related to health. According to social support theory, receiving support from others is generally beneficial to mental and physical health, and it may blunt the harmful impact of stressful experiences, including the disease itself.[40] Empirical confirmation of this "buffering hypothesis of social support" has been found among patients with various rheumatic diseases.[41] Irrespective of the disease, the presence of a spouse or intimate partner, having many close social relationships, being socially active, and perceiving others being supportive have been found to have favourable effects on psychological and physical functioning.[42]

### 5.1 Social activities

Illnesses such as EDS typically have various social consequences (Box 20-2). In a Dutch survey, five times as many patients with EDS reported problems with social functioning than did people from the general population, and the chance of experiencing problems with role functioning due to physical problems was seven times as large as in the general population (figure 20-1). Interview studies also show that impaired physical functioning in EDS hampers social life.[8,32]

**Box 20-2** Possible social consequences of having a rare chronic disease

- Problems with work and pursuing social activities.
- A change of role patterns, household tasks, and leisure activities in the family; difficulty to decide to get offspring; difficulty deciding on the way to raise children with EDS.
- Sexual problems.
- Problems due to lack of knowledge of health care workers.
- Experiencing lack of understanding and acknowledgement.

### 5.2 Family

Having a chronic disease affects one's family. For example, the kind and number of leisure activities and roles and household tasks of family members often change as a consequence of illness. Yet, not all consequences are negative. A clinical consultation with a man with EDS revealed that he had to quit his work due to EDS, but he stayed at home and did household tasks, which also allowed him the privilege of seeing his children more frequently.

Parents of children with EDS may be conflicted between protecting their children as much as possible, or encouraging them to face the challenges of life and not be hindered by fear of joint dislocations and other EDS consequences.[8] Moreover, parents need to decide when and how they will inform their children about EDS, which is especially difficult for parents of children with vascular EDS. The decision about whether to have children in the first place and when so, whether or not to pursue prenatal diagnosis can be very difficult. During a

pregnancy, worry over the foetus is common. If the child does have EDS, guilt among parents may occur.

### 5.3 Sexuality

Sexuality is closely related to other dimensions of quality of life. The sexual life of patients with EDS may be hampered by pain, fear for damage to the vaginal tissues, and the need to avoid certain positions because of the risk of joint dislocation and pain.[8] In a small and non-representative sample, 60% of the women with EDS reported experiencing pain during sexual intercourse.[43] Pain, fatigue, and depressed mood can impair the various phases of the sexual response cycle: desire, excitement, plateau, orgasm, and resolution. Ideally, patients and their partners should discuss sexual problems and mutually find behaviours and positions that reduce pain or risk. Such communication is difficult for many couples, however, and consultation from a physician or a psychologist may be helpful.

### 5.4 Health care

Patients with EDS may experience problems with both the lack of recognition and lack of knowledge of EDS by health care providers.[44] The disease may be diagnosed only after a long time of fruitless contacts with physicians and other health care workers. Patients may have the feeling that others suspect them of hypochondria or simply that they are complaining too much. Patients may feel that they have to educate health care providers about EDS, rather than the other way around.[8] Patients with EDS were interviewed about their health care experiences, and five categories of problems were identified: being ignored and belittled by health-care professionals, being assigned psychological or psychiatric explanations, being treated and considered merely as an object, being intruded in one's personal sphere when care is given, and being suspected of family violence.[45] Unfortunately, the consequences of these experiences were that patients did not fully trust their physicians, which risked increasing their health problems. These authors advised health care professionals to base their actions on norms that protect human dignity and confirm each patient as a unique human being with resources and abilities to master his or her own life.

### 5.5 Society

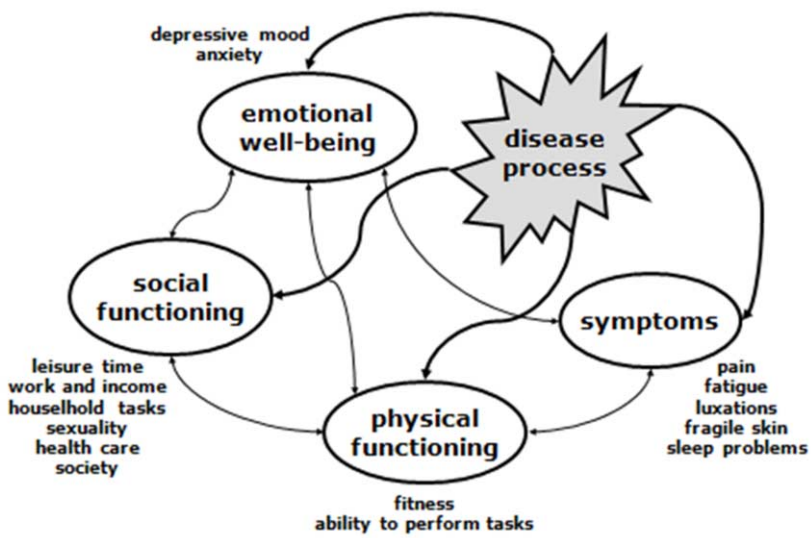
These interviews of patients with EDS also revealed that they experienced a lack of understanding from society at large.[5] For example, people may experience shame because they use a wheelchair when other people do not understand the need for it (“You can walk, but you use a wheelchair?”). Patients must often demonstrate great perseverance to obtain the accommodations or aids that they need, such as a disabled parking permit or disability status. Because symptoms such as pain, fatigue, and the chance of joint dislocations are not visible, it can be problematic to have to repeatedly explain that is difficult to perform specific tasks at work. Prevention of social isolation and social problems is a shared responsibility of the patient with EDS and the people who are in contact with the patient. Fellow-sufferers of EDS can be of great support.

## 6. Conclusion

There are few studies of psychosocial factors in EDS. Moreover, those studies that are available are often in small and non-representative samples. Nevertheless, the data uniformly suggest that reduced quality of life is common. Fatigue, pain, joint dislocations, skin problems, ‘weak’ vessels and organs, and sleeping disturbances threaten both psychological well-being and physical and social functioning. Yet, the effects of EDS on quality of life differ widely among individuals. Although some people with EDS have serious disease and poor quality of life, others are able to live a happy and satisfying life despite the serious

disease, and for still others the disease is milder and self-care and quality of life are good. Furthermore, improvements in one aspect of quality of life (i.e. symptoms, emotional well-being, physical, or social functioning) often yield favourable consequences for other aspects (figure 20-2). Coping abilities and social support can buffer the adverse consequences of the disease. For a patient with severe EDS, a single physician as case manager should coordinate care. This case manager knows the medical file of the patient and can help the patient to decide about medical, psychological, or occupational therapies. Patients who are having trouble adapting successfully to their condition, should be encouraged to seek behavioural help, especially from someone trained in cognitive-behavioural interventions. Health care workers, patient associations, and people in the close environment of patients can help them find the appropriate education, counselling, and multidisciplinary interventions aimed at dealing with the adverse consequences of EDS.

**Figure 20-2** The consequences of a chronic disease such as Ehlers-Danlos syndrome for various mutually dependent aspects of well-being and functioning




*The disease process and its symptoms threaten emotional well-being and physical and social functioning. Yet, the effects of Ehlers-Danlos syndrome on quality of life differ widely among individuals. Although some people have serious disease and poor life quality, others are able to life a happy and satisfying life despite the serious disease, and for still others the disease is milder and self-care and life quality are good. Furthermore, improvement in one aspect of life quality (i.e., symptoms, emotional well-being, physical, or social functioning) often yield favourable consequences for other aspects. Coping abilities and social support can buffer the adverse consequences of the disease.*

**7. Areas of uncertainty**

Studies of somatic problems, psychological distress, and social functioning in EDS are scarce, but quite consistent in the conclusion that quality of life is, on average, impaired in these domains. Our discussion and recommendation in the area of coping and therapeutic possibilities were almost fully based on our knowledge of experiences of individual patients, and our knowledge of the somatic, emotional, and social consequences of chronic disease

more generally. Disease-specific knowledge, especially on the effects of cognitive-behavioural interventions, is urgently needed.

## 8. Summary

This chapter describes the somatic, psychological, and social impact of EDS. Research indicates that pain and fatigue are common problems in EDS. In addition, patients may fear damage to their joints, skin, and other organs; progressive disability; and even death. Other psychological and social dilemmas or challenges often occur, including whether or not to have a child, finding or keeping work that is commensurate with abilities and interests, altered roles in the family, a lack of understanding by others in one's social environment, and obtaining appropriate health care. Yet patients vary widely with respect to the severity of the disease and its impact on their psychological, social, and physical functioning and well-being; individual assessment is needed. It is probably optimal to have one physician manage each patient and coordinate the many aspects of his or her care. Patients should have access to education, counselling, and multidisciplinary interventions aimed at reducing the adverse consequences of the disease.

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