ARVC-Selbsthilfe Patient Survey for ARVC patients and relatives

Subject

Needs of patients when being diagnosed with ARVC

Period

Start: December 2019
Ongoing (Status: 01.12.2020)

Response

Participants	**26**
Patients with symptoms / diagnostic criteria	23
probands (first patient in family diagnosed)	19
identified via family screening	4
Mutation carriers without symptoms / criteria	0
Family member without ARVC / ARVC mutation	3

Questions asked

When confronted with the diagnosis of ARVC:

1. What kind of questions have arisen at this moment?
   What information would have been necessary?
2. What needs were there? Were they fulfilled?
3. Which media should be used to support you?
4. Would individual or group counselling have been necessary?
   If so, on which topics would individual counselling have been preferable?
   If so, on which subject areas would group counselling have been sufficient?
5. What direct effects does the disease have on your everyday life?
6. Have you been offered psychological support?
   If so, at what time? Have you accepted it?
   If not: would you have liked to have it and if so, at what time?

Results of the survey

(in italics: verbatim quotes from the survey)

1. What kind of questions have arisen at this moment? What information would have been necessary?

a) Medical issues:

- **Basics**
  “What is ARVC?”
- **Symptoms**
  What are the possible symptoms?
  “Which of my symptoms are normal /abnormal / dangerous?”
- **Reliability of the diagnosis**
  “Can this be a misdiagnosis?”
  "Why was this diagnosis missed for so long?" (after 10 years of diagnostic odyssey)
- **Risk stratification**
  “Will I need an ICD?”
- **Possible therapies (medication, ablation, ICD)**
  side effects, risks, benefits of therapeutic treatments
  decision for/against offered therapies
  “Will a transplant be necessary in the end?”
- **Next steps**
  frequency of monitoring, required therapeutic measurements, medical care
  “Where are the ARVC specialists (clinical, outpatient)?”
  “Are there specialized psychocardiologists?”
- **Curability**
  “If there is no cure – what about research?”
- **Prognosis**
  “What does the future look like?”
  “Will the situation worsen, stay the same or improve – compared to now?”
- **Inheritance**
  “Are more relatives affected?”
  “What does this mean for my children?”

b) Social/practical issues

- **Job / Work**
  “Can I continue in my old demanding job?”
  “Will I be able to work full-time again?”
  “Will I get a job at all with this disease?”
  “Who is going to pay for a retraining?”
- **Rehab**
  “Are there specialized rehab centres for ARVC patients?”
- **Disability**
  “I don´t feel handicapped? Am I nevertheless entitled to a pass for severely disabled persons?”

- **Insurance**
  "Will I be able to get a private health insurance?"
  "Will I be able to get a life insurance to provide financial security for my relatives if I die of sudden cardiac death?"
- **Money**
  "Who is going to pay for the genetic testing (of me/ of my deceased relative)?"
  "Will I be able to pay a psychotherapy?"

c) Lifestyle issues

- **Sports / exercise**
  "I've been thinking about every little walk, every staircase climb, every bike ride if I could do something "wrong" and would damage my heart"
  "How much exercise is possible or harmful?"
  "What does: No sports mean? What about walking, jogging, cycling, yoga, fitness?"
- **Beneficial lifestyle factors**
  "What can I do myself to consolidate my condition?"
  "What are the factors influencing the disease?"
  "How can I positively influence the disease? What do I have to change?"
- **Traveling**
  Worries about traveling in countries with bad health system, in tropical regions (because of the climate), in height (because of oxygen deficiency)
- **Family planning**
  "Will I pass this mutation on to my children?"
  "Will I get the permission for preimplantation diagnostics?"
  "Can I find a partner who accepts a sick partner and the possibility of passing on the mutation?

d) Existential questions

- **Quality of life**
  "What does this mean for my everyday life – in the short, medium and long term?"
  "What quality of life remains for me?"
  "What restrictions do I have to live with?"
  "Can I still participate in life? Am I forever different?"
  "Basically, I felt like my life was over!"
- **Life expectancy**
  "Must I die?"
  "Will I die early?" (girl, 16 yrs)
  "Will I still be there for my children as long as they need me?" (Mother, 33 yrs)
- **Family conflicts**
  Different approach and handling of the disease in the family (with regard to genetic testing, openness, talking about the disease)
  blaming family members
  "It's my mother-in-law's fault because she passed the gene mutation on to my wife and kids"
- **Individual conflicts**
  "Should I get tested?" (family member of proband, 32 yrs)

- **Coping**
  "How can I go on living with this disease?"
  "How can I as a mother bear this and learn to live with it?" (Mother, 66 yrs)
  "How can I find others who are going through the same thing?"
  "Who will help me learning to live with this disease?"
  "Who tells me about stabilizing factors?"
  "Where can I find support? Are there specialised psychologists? Are there self-help groups?"
  "Who can give me orientation in chaos?"

e) Emotions

- **Anxiety / fear**
  Fear of death
  "Am I going to die?"
  Fear of losing a loved one
  "Will anyone else in my family have to die of this disease?"
  Fear of cardiac arrhythmia and ICD shock
  "I hardly dare leave the house for fear of the next ICD-shock"
  Fear of being abused as guinea pig
- **Sadness / Depression**
  "I had to give up my favourite hobby" (sportive man, 28 yrs)
  "I have lost nearly all of my friends"
- **Anger**
  "Why does this happen to me of all people?"
  But also: *"I don´t want pity"*
- **Denial /Refusal / Repression**
  "I did not want to believe that I am ill" (athlete, 22 yrs)
- **Overstrain**
  "I have no idea how to manage"
- **Pain / grief**
  "Will this pain last forever?" (mother, 52 yrs, after loss of her son)
- **Guilt**
  "Why did I have to pass on this terrible disease to my children?"
- **Other**
  Near death experiences during resuscitation / aborted cardiac arrest

2. What needs were there?
Were they fulfilled?

Yes	3	12 %
Partly	5	19 %
No	18	**69 %**

a) **Needs met (3 out of 26)**

The 12% indicating that their needs were met, were patients with medical background (themselves or in their family) or patients with knowledge about the disease because there was already an affected family member. They were satisfied with the medical information because they already knew about the disease or because they understood well because of their background.

b) **Needs partly met (5 out of 26)**

Patients indicating that their needs were only partially met were satisfied with the answers to their medical questions but dissatisfied with the psychosocial support provided

c) Needs not met (18 out of 26)

Over two thirds of the patients told us that their needs were not met at all.

What was missing?

- **Lack of expertise**
  diagnostic odyssey (sometimes more than 10 years)
  "My doctor had never seen an ARVC patient until now"
  Misinterpretation of arrhythmias as mental problems
  "I was sent to a psychiatrist to solve my problems"
  "I have been told that I am mentally ill"
  "Possible differential diagnoses were not considered"

- **Lack of time**
  no time for explanations, discussions, pros and cons (benefit-risk assessment)
  not enough time for important decisions
  "I was supposed to decide on the type of ICD within a day"

- **Lack of information**
  no answers to urgent questions
  Lack of comprehensible explanations (too many technical terms)
  "I didn´t understand what the doctor was talking about"
  no possibility of informed consent
  no reference to sources of information (like a brochure or the link to a website)

- **Lack of organisation**
  every time another doctor / geneticist
  “I had to tell my story over and over again”
  long waiting periods for appointment (physician/psychologist)
  hours of waiting for procedures
  months of waiting for results
  Young patients together in the room of a hospital with old seriously ill patients (being resuscitated/dying)

- **Difficulties in doctor-patient relationship**
  lack of understanding and respect by the doctor for individual decisions of the patient
  lack of interest of the doctor in the patient's circumstances and individual situation
  “The doctor wasn't interested in me at all!"
  Ignoring the psychosocial needs of the patient
  "There were only medical answers to our most terrified questions."
  Denial of genetic testing on children
  "I wanted to know if my children were also affected by the disease. But the doctor didn't do the genetic test – he told me the children were still too young”
  lack of trusting relationship
  "The doctor didn't tell me the whole truth!"
  Overload the doctor by the demands of the patient
  "I was a teenager, the doctor was slightly overstrained with a teenager and could not respond adequately to my refusal" (young woman, 22 yrs, diagnosed with 16 yrs)

- **Lack of support**
  Lack of encouragement
  "I would have liked the doctor to encourage me that it is possible to live well with the disease"
  “Nobody gave me security, nobody encouraged me to say yes to life despite the threat of sudden cardiac death”
  No psychosocial support options / psychosocial care
  "There were no offers of help - only the question: Will you be all right?"
  "Why was my relative sent home without any offer of help?"
  “Nobody explained to me about the interaction of heart and soul.”
  no contact to other patients, patient forums or patient organisations
  "Reference to a support group or patient forum would have been helpful."
  "The doctor said that he was not allowed to put me in touch with another patient for privacy reasons."
  Lack of support for depression and suicidal phases
  "I probably would have needed psychological support at that point."
  “Nobody told me how to overcome my fears”
  "I felt completely left alone!"

3. **Which media should be used to support them?**
    - Internet
    - Patient brochure, information material to take away
    - Awareness campaigns through television, radio, print media
    - Animation to illustrate the mechanisms of the disease
    - YouTube Videos
    - FAQ with explanations that can be understood by patients
    - e-mail, newsletters
    - Self-help groups
    - patient forums, contact with other fellow sufferers
    - Testimonials from other patients
    - Peer group consulting
    - Online Q&A with experts, video chat
    - Telemedical services (digital examination results, avoiding long journeys)
    - List of psychologists/psychotherapists

4. **Would individual or group counselling have been necessary?**

Yes	22	85 %
No	3	11 %
Not specified	1	4 %

a) If yes: on which topics would *individual counselling* have been preferable?

- medical topics such as initial diagnosis, classification of symptoms and complications, individual characteristics and risk, individual therapy recommendations, prognosis
- Family conflicts
- Psychosocial issues
- individual coping
- individual stabilizing factors
- traumatic experiences
- individual recommendations for exercise and sports
- individual recommendations for family members
- Questions on quality of life and life expectancy
- Psychotherapy

b) If yes: on which topics would *group counselling* have been sufficient?

- General educational topics, general medical questions
- Exchange on the organisation of everyday life / dealing with the illness / integration into life
- Relaxation methods
- social issues
- exchange of experiences among patients
- Peer group counselling e.g. from young people for young people (on sports)
- family education
- Group therapy
- Innovations and new treatment methods

5. **What direct effects does the disease have on your everyday life?**
    - Limited physical capacity → Priority setting necessary
    - Mental stress
    - concentration disorders, fatigue
    - rigid daily structure, sometimes incompatible with "normal" social life
    - hot phases not easy to plan → difficult for social contacts
    - no job → time must be filled and structured differently
    - Learning process (necessity of abandoning parts of the previous life)
    - Anxiety, especially in the case of spontaneous and increased cardiac arrhythmias
    - permanent fear and concern for relatives
    - Abandoning activities and social events
    - Poor stress resistance at work and at home
    - Sleep disturbances (because of arrhythmia or anxiety)
    - renouncing dear hobbies (sports...)
    - Loss of friends
    - Conflicts with partner and family
    - Restrictions in dealing with children and grandchildren
    - Spending a lot of time in hospitals and outpatient clinics
    - Special restrictions due to corona (social isolation)
    - Loss of confidence in one's own body (what is still possible?)
    - Fears about the future
    - Loss of life plan (training/university/job)
    - Side effects of medication
    - loss of working capacity, early retirement
    - restricted traveling
    - limited mobility (no driving)
    - abstention from alcohol
    - Insecurity (what can/may I do? What is too much?)

    Positive aspects
    - In the long run: more calmness, more balance
    - concentration on the essential issues of life
    - appreciate life as precious
    - more profundity in life

6. Have you been offered psychological support?

Yes	4	15 %
No	22	**85 %**

Apparently there is a gap between expert centres which offer support in 48% of cases (as indicated in the ERN GUARD-Heart pre PROMS Online Survey), and other health care providers. **85% of the patients have not been offered psychosocial support at all.**

a) You answered "No, I have not been offered support" (22 out of 26)
Would you have liked support?

Yes	15	**68 %**
Probably	3	14 %
No	4	18 %

You answered "Yes, I would have liked support": at what point/in which situation?

- directly after diagnosis
- 4 – 6 weeks after diagnosis
- after ICD implantation (directly / 4 – 8 weeks later)
- If symptoms worsen
- when children are tested positive for the mutation

More than two thirds of the patients would have appreciated psychological help.

b) You answered "Yes, I have been offered support" (4 out of 26)
If so, at what time?

- Directly after diagnosis
- after ICD implantation
- years after the diagnosis when problems were apparent

The patients who were offered support were in hospital after dramatic events or for ICD implantation. In one case the general practitioner suggested the psychological support. Nobody was offered support in an outpatient clinic.

Have you accepted it?

Yes	3	75 %
No	1	15 %

Problems identified

- Find a sympathetic psychologist who meets my needs
- Long waiting periods for psychotherapy

Conclusions from the survey

What needs to be done in the future from the patient´s perspective?

- Psychologist should be part of multidisciplinary team in inherited cardiac clinic unit
- psychosocial screening via questionnaires to assess patients´ needs should be used
  Patients may appear calm on the outside, but are very vulnerable on the inside
- adequate time for information and shared decision making should be provided
- psychosocial support should be offered to every patient
  early intervention can prevent chronification of mental health problems
- continuity in patient follow-up should be ensured for a trusting doctor-patient relationship
  the stories of patients are so complex that they cannot be briefly summarised for a new doctor, genetic counselor or psychologist
- information on further support options should be provided
  collection of a list of psychologists, therapists, patient forums and patient organisations
- psychosocial guidelines / recommendations for inherited cardiac diseases are necessary