Welcome

ERN GUARD-Heart
PROMS - PREMS Workshop

Working patient-centred in the context of cardio-genetics

Module 2: Patient-centric approach

Tuesday, 17 November 2020

European Patient Advocacy Group

ARVC SELBSTHILFE

European Reference Network
for rare or low prevalence complex diseases
Network Heart Diseases (ERN GUARD-HEART)

Introduction

Dr. Ruth Biller

ARVC-Selbsthilfe e.V. Board Members

Gynaecologist

ARVC-Selbsthilfe e.V. (Chair)

European patient advocacy group ePAG of ERN GUARD-Heart (Chair)

Verwaiste Eltern und trauernde Geschwister München e.V. (Compassionate Friends, Support of bereaved parents, Counselling after Sudden Cardiac Death)

Dr. med. Ruth Biller

Ärztin und Trauerbegleiterin

Schwerpunkt: Plötzlicher Herztod

ARVC-Selbsthilfe e.V.

2013 founded as self-help group **ARVC-Selbsthilfe**

2018 recognised as Non-profit Organisation **ARVC-Selbsthilfe e.V.**
European patient advocacy group (**ePAG**) ERN GUARD-Heart

European Patient Advocacy Group

European Reference Network
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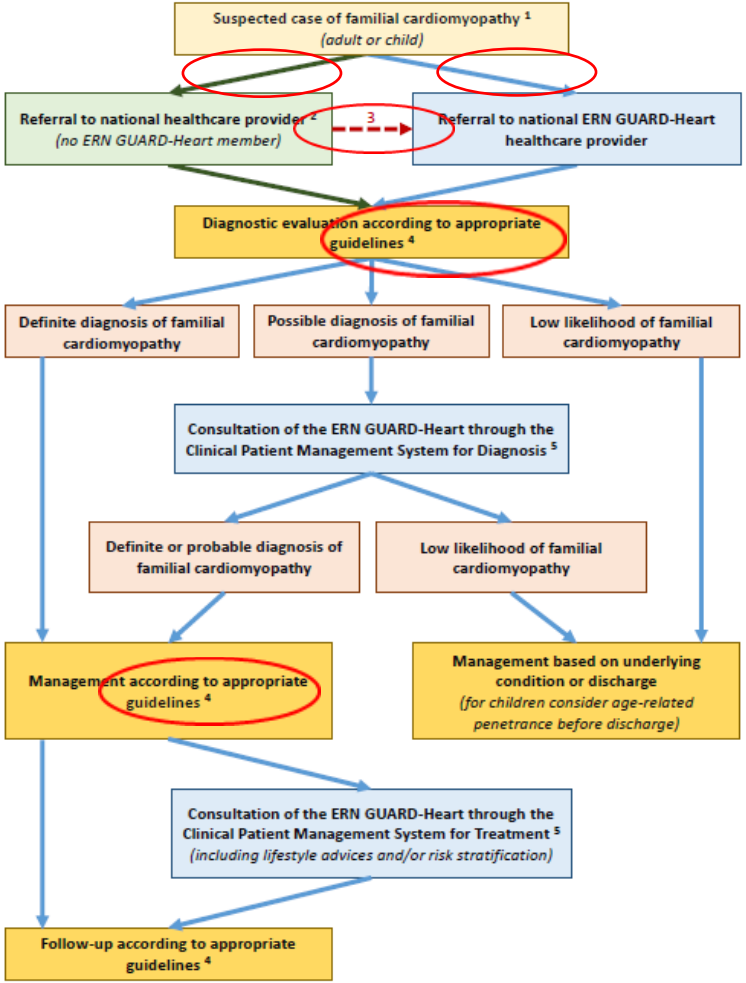
2020
132 members (82 ARVC patients / mutation carriers)
250 active users of our services
38.000 visitors / year to our website

Aims and activities
https://guardheart.ern-net.eu/patients/epags/arvc-selbsthilfe/

New initiatives in 2020
https://www.arvc-selbsthilfe.org/english/

ARVC SELBSTHILFE

ERN GUARD-HEART CROSS-BORDER PATIENT PATHWAY FOR FAMILIAL CARDIOMYOPATHIES



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Footnotes

1. Primary care (general physicians) or secondary care (treating physicians in district general hospitals).
2. If possible, healthcare provider with an Inherited Cardiac Clinic Unit with a multidisciplinary team.
3. Consider, if possible, referral to a national ERN GUARD-Heart healthcare provider (before or after the diagnostic evaluation process).
4. See list of appropriate guidelines (next page).
5. See *Quick user Manual for CPMS* or contact the management office of ERN GUARD-Heart for support through the official website of ERN GUARD-Heart (http://guardheart.ern-net.eu/about/contact/).

European Reference Network

ARVC – arrhythmogenic right ventricular cardiomyopathy Patient Journey

1. Pre-diagnosis
Apparently healthy person without any symptoms or only slight symptoms which may be ignored.

2. Symptoms
(Exercise related) palpitations, tachycardia, premature ventricular contractions (heart stumbling), arrhythmias, atypical breast pain, pre-syncope (dizziness, light-headedness), syncope (fainting), dyspnea (shortness of breath).
In extreme: sudden cardiac death (SCD) due to ventricular fibrillation (VF) or cardiac arrest

3. Diagnosis
Clinical history
Physical examination
Family history (detailed!)
12-lead-ECG
Exercise ECG
Ambulatory ECG monitoring
Echocardiography
Cardiac MRI
Endomyocardial biopsy?
Electro-physiological study?
Genetic testing?
Implantable loop recorder?

4. First treatment
Medicines (antiarrhythmic drugs like beta-blockers, sotalol, amiodarone, flecainide / drugs for heart failure);
ICD in case of high risk for SCD;
Catheter ablation to treat arrhythmias

5. Surgery
Heart transplant only if severe heart failure or failure of drug therapy/ablation with high load of arrhythmias

6. Follow-Up Care
Close cardiac check-up (at least once a year, with ICD normally twice a year, more often if high risk situation)
Regular risk stratification to re-evaluate need of ICD

7. Psychological Care / Rehabilitation / Social Care
Anxiety (ICD-shocks, frightening arrhythmias, death…)
Deep grief in case of SCD of a relative

1. Note: in case of SCD without any preceding warning symptoms: autopsy and genetic testing.

1. Ideally: Be aware of first warning symptoms.
In discussion: genetic screening of top athletes.

2. Note: Heart symptoms especially in young people should arouse suspicion.
Ask for cases of unexplained SCD in the family history.

2. Ideally: Complete cardiac check-up including ECG, Ultrasound, MRI
Transfer to specialist to get the correct diagnosis, if necessary.
Genetic testing to secure diagnosis.

3. Note: Identify endangered patients (risk stratification). Need of information about the disease and implications for relatives. Find psychological support nearby and without long waiting period

3. Ideally: Risk stratification in dialogue between patient and doctor. Identify affected relatives by genetic testing (cascade family screening) and/or cardiac check-up, special advice concerning genetic testing of children.
Chance to do all the analysis in the same place. Transfer to specialist if necessary. Sufficient time for questions and advice.

4. Note: Side effects of medication
Advice concerning lifestyle, physical activity, sports, travelling, personal risks…
4. Ideally: Better guidelines to identify endangered patients
Sufficient time for questions and advice. Transfer to specialist if necessary

5. Note: Find psychological support nearby and without long waiting period
5. Ideally: Transfer to transplantation expert center

6. Ideally: Identify optimal time to implant an ICD to prevent SCD.
Transfer to specialist if necessary.
Advice for family planning (50-50 risk if gene identified). Find a cardiologist nearby who is willing to do the follow-up but transfers to specialist, if necessary.
Ideally always the same contact person.

7. Note: Find psychological support nearby and without long waiting period
7. Ideally: Good psychosocial care
Rehabilitation units who know how to deal with ARVC patients. Advice for insurance/work/driving/card for severely handicapped.

ARVC-Selbsthilfe Patient Survey (Dec. 2019 – Nov. 2020)

What were we asking for?

The needs of patients when diagnosed with ARVC

How many responded?

25 participants

Patients with symptoms / diagnostic criteria: 22
- Probands: 19
- Patients identified via family screening: 3

Mutation carriers without symptoms / criteria: 0

Family member without ARVC / mutation: 3

Relevance

This survey is biased:

- Patients who turn to a self-help organisation have a special need for support
- Patients who responded to the survey have even stronger concerns

Nevertheless the survey shows existential needs of many patients

ARVC-Selbsthilfe Patient Survey
about needs of patients when being diagnosed with ARVC

Questions asked

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?

ARVC-Selbsthilfe Patient Survey

1.+5. Arising questions, necessary information, impact on everyday life

a) Medical questions

- Information on the disease

b) Social issues

- Job / Work
- Rehab
- Disability
- Insurance
- Money

c) Lifestyle issues

- Sports / exercise
- Beneficial lifestyle factors
- Traveling
- Family planning

ARVC-Selbsthilfe Patient Survey

1.+5. Arising questions, necessary information, impact on everyday life

d) Existential questions

- Quality of life
  “Basically, I felt like my life was over”
- Life expectancy
  “Will I die early?” (girl, 16 years)
- Family conflicts
  “It's my mother-in-law's fault because she passed the gene mutation on to my wife and kids”
- Coping
  “How can I go on living with this disease?”

ARVC-Selbsthilfe Patient Survey

1.+5. Arising questions, necessary information, impact on everyday life

e) Emotions

- Anxiety / fear
  “I hardly dare leave the house for fear of the next ICD-shock”
- Sadness
  “I had to give up my favourite hobby” (sportive man, 28)
- Anger
  “Why does this happen to me of all people?”
- Denial
  “I did not want to believe that I am ill” (athlete, 22)
- Pain / grief
  “Will this pain last forever?” (mother, 52, after loss of her son)
- Guilt
  “Why did I have to pass on this terrible disease to my children?”

ARVC-Selbsthilfe Patient Survey

Question 2: What needs were there? Were they fulfilled?

Were the patient´s needs met?

- Yes: 3 (12%)
- Partly: 5 (20%)
- No: 17 (**68%**)

Needs not met – what was missing? (1)

- **Lack of expertise**
  diagnostic odyssey
  “My doctor had never seen an ARVC patient before“
  Misinterpretation of arrhythmias as mental problems
  “I was sent to a psychiatrist“
- **Lack of time**
  no time for explanations, discussions, pros and cons
  “I was supposed to decide on the type of ICD within a day”

ARVC-Selbsthilfe Patient Survey

Needs not met – what was missing? (2)

- **Lack of information**
  no or incomprehensible answers to urgent questions
  “I didn´t understand what the doctor was talking about”
- **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
- **Difficulties in doctor-patient relationship**
  lack of understanding individual decisions
  “The doctor showed no interest at all”
- **Lack of support**
  No psychosocial support options
  “I felt completely left alone”

ARVC-Selbsthilfe Patient Survey

Question 6: Have you been offered psychological support?

- Yes: 3 (12%)
- No: 22 (88%)

If no: would you have liked support?

- Yes: 15 (68%)
- Probably: 3 (14%)
- No: 4 (18%)

And at what point?

- 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

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

- Psychologist part of multidisciplinary team in inherited cardiac clinic unit
- psychosocial screening via questionnaires to assess patients´ needs
- Provide adequate time for information and shared decision making
- Offer psychosocial support to every patient
- Ensure continuity in patient follow-up for a trusting doctor-patient relationship
- Provide information on further support options
- psychosocial guidelines / recommendations for inherited cardiac diseases

Our common goal:

Trusting cooperation between doctors, geneticists, psychologists, social carers, patients and patient organisations

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Please use the resources and expertise of patient organisations

They provide

- Expertise of living with the disease
- Knowledge transfer
- Closeness and Warmth
- Community and Exchange
- Solidarity and Encouragement
- Solutions for everyday problems

Thank you!

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