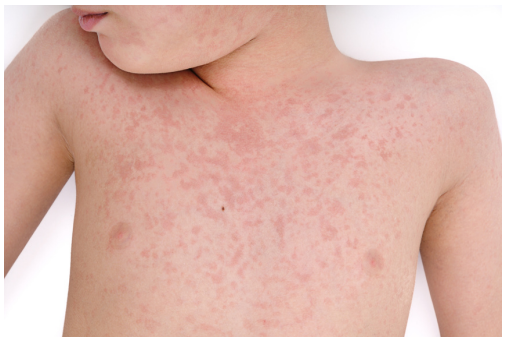
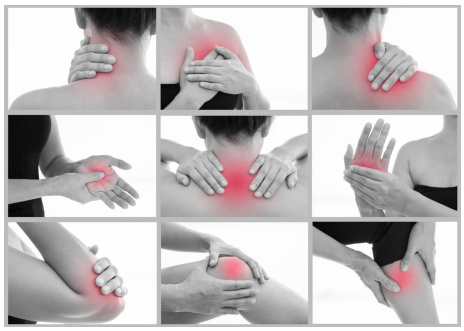
RARE AUTOINFLAMMATORY CONDITIONS COMMUNITY - UK

BEING A VOICE FOR PATIENTS, FAMILIES AND CARERS LIVING WITH AN AUTOINFLAMMATORY CONDITION








PATIENT HANDBOOK

REGISTERED CHARITY IN ENGLAND AND WALES 1184846

RACC-UK

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About this handbook

This handbook is created as a response to the Governments 'Rare Disease Framework' across the UK. One recommendation made was for patients to have a Medical Alert Card to help assist their care in cases of emergencies.

As patients, parents and carers become their own coordinator of their own care, and because some systems within the NHS may not be linked to each other (e.g. cross regions), this handbook will help you to keep all of the relevant information in one place.

You can make several copies of this handbook if you wish. It may be worth giving school / college or even your employer a copy to keep on file so that they know more information about you and your condition.

England Rare Disease Action Plan (2023)
(https://www.gov.uk/government/publications/england-rare-diseases-action-plan-2023)

Scotland Rare disease action plan (2022)
(https://www.gov.scot/publications/rare-disease-action-plan/)

Wales Rare Disease Action Plan (2022 to 2026)
(https://www.gov.wales/wales-rare-diseases-action-plan-2022-2026-whc2022017)

Northern Ireland Rare Disease Action Plan (2022 - 2023)
(https://www.health-ni.gov.uk/sites/default/files/publications/health/doh-ni-rare-diseases-action-plan-2223.pdf)

About RACC - UK

Our Vision

- For all UK patients, directly or via parents and carers, to have genetic testing and reach a diagnosis for Autoinflammatory Conditions.
- For UK patients, parents, and carers to be able to access the right treatment through the NHS
- For the public, carers, and medical professionals to be well educated about both the emotional and physical effects of Autoinflammatory Conditions.
- For all patient care to be well coordinated
- For all patients, parents, and carers to be able to access relevant Disability/ Sickness benefits for financial support such as Disability Living Allowance/ Personal Independence Payment, Employment and Support Allowance, and Carers Allowance.

What do we do?

- Provide Peer Support
- Provide 1:1 Support Meetings (Phone or Zoom)
- Write Supporting Letters
- Policy - For example, we work with NHS as stakeholders to inform policy through drug consultations. We provide patient impact statements to ensure the patient experience is accurate.
- Work alongside Medical Professionals
- Educate and raise awareness with schools
- Liaise with Housing Associations

How can we help?

- We can arrange 1:1 meetings with you and create an action plan to help you manage challenging times.

With your signed consent we can also liaise with:

- medical teams,
- school,
- employers,
- housing associations.

(+44) 07377091552

support@raccuk.com
www.raccuk.com

About Me

Full Name
Address

DOB (DD/MM/YYYY)
NHS Number:

Diagnosis	Genetic Mutations (if known)	Year Diagnosed

My picture

In Case of Emergency Please Contact:

NAME:

Relationship:

Phone Number:

Address:

(+44) 07377091552

support@raccuk.com
www.raccuk.com

My likes and dislikes

(Please edit for your own preferences)

My favourite food is	Food
I like watching	TV Program / Film
I like playing....	Sports
I like playing.....	Games (PC / XBOX / PS/ SWITCH / BOARD GAMES / CARD GAMES)
My favourite teddy / toy is....	Teddy / Toy Name
I dislike	Food
I dislike	Blood Tests
I dislike	Decisions being made without my involvement
I dislike	Feeling poorly and sometimes I get scared
I dislike	People touching me without asking

Medications

MEDICATION	DOSEAGE	FREQUENCY

(+44) 07377091552

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Symptoms

- **Fevers**
- **Rigours (shakes with fevers)**
- **Sore Throats**
- **Gastrointestinal Issues**
- **Enlarged Spleen and Liver**
- **Genital Ulcers**
- **Inflammation of the joints and skin**
- **Uveitis (inflammation in the eyes)**
- **Respiratory Issues**
- **Headaches**
- **High C - Reactive Protein (CRP)**
- **Enlarged Lymph Nodes (Glands)**
- **Constipation**
- **Nausea**
- **Diarrhoea**
- **Rashes**
- **Anaemia**

Autoinflammatory conditions, otherwise known as Periodic Fever Syndromes, are not to be confused with Autoimmune conditions such as Rheumatoid Arthritis or Crohn's Disease.
Autoinflammatory conditions are rare and complex mainly caused by changes in genes. Autoinflammatory conditions are associated to the innate immune response, where as Autoimmune conditions are associated to the adaptive immune response.

[Autoinflammatory Syndromes | Centre for Amyloidosis and Acute Phase Proteins - UCL – University College London]

Blood Test Information

Disclaimer: The following information has been approved by our Medical Advisory Board but please discuss your results with your GP or Consultant. This is an information guide about what each test is for, not a diagnostic tool.

Blood tests are often performed regularly on patients with diagnosed or suspected autoinflammatory disease. The results can be used to:

· Aid diagnosis
· Look for signs of inflammation, for example spikes of inflammation during flares
· Determine if treatment being given is working effectively
· Check for 'red flags' requiring urgent treatment, such as excessive immunosuppression if on treatment

Blood tests commonly performed for autoinflammatory disease patients include:
· Genetic testing
· C-Reactive Protein (CRP)
· Erythrocyte Sedimentation Rate (ESR)
· Serum Amyloid A (SAA)
· Full blood count
· Amongst others such as general tests of liver and kidney function

C-Reactive Protein (CRP)
CRP is a protein which is produced by the liver and found in blood plasma. The levels are usually very low in a healthy individual, and often not detectable. CRP is used as a marker for inflammation in the body. CRP is often raised in autoinflammatory patients experiencing a flare of their symptoms and can be slightly raised outside of a flare. Other causes of raised CRP can include infection or as a reaction to physical trauma.

Erythrocyte Sedimentation Rate (ESR)
ESR is a method used to monitor inflammation within the body. Like CRP, it does not give a specific diagnosis or cause of symptoms but can evidence inflammation within the body. The test involves measuring the time it takes for a red blood cells (erythrocytes) to fall to the bottom of a tube of blood. In individuals with increased inflammation, the red blood cells will fall more quickly, which will give a higher ESR result. Other causes of raised ESR include infection, anaemia, pregnancy and old age.

Serum Amyloid A (SAA)
SAA is another inflammatory biomarker which can be monitored in individuals with suspected autoinflammatory disease. SAA is a protein which is released in response to inflammation in the body. The levels of SAA are also monitored to assess the effectiveness of an individual's treatment, and to identify the risk of long-term complications such as amyloidosis. If SAA remains high even whilst on treatment, it is an indication that higher dosage or alternative treatment may be required. A high SAA does not equate to systemic amyloidosis, however.

Full Blood Count (FBC)
The full blood count is common test that looks at a patient's overall health. It looks at a patient's red blood cells, haemoglobin, white blood cells and platelets.

Red blood cell count shows the number of red blood cells present in a volume of blood. Low red blood cell count and haemoglobin levels could indicate anaemia. Haemoglobin count looks at the haemoglobin content of the blood. This is a protein which is found in red blood cells, it is what carries oxygen in the blood and causes blood to be red in colour. Low levels indicate anaemia.

Mean Corpuscular Volume (MCV) is the average (i.e. mean) volume of red cells within a blood specimen. This test's value is low if red cells are small, i.e. microcytic, or high if the red cells are large, i.e. macrocytic. The size of red cells helps doctors determine the possible cause of anaemia, when considered together with other blood levels such as 'Hb' or Haemoglobin.

White blood cell (WBC) count is the number of white blood cells present in a volume of blood. Low white blood cell count can indicate immunosuppression (reduction in the effectiveness of the body's immune system), possibly due to various treatments used to treat autoinflammatory disease. The most common cause of a high white blood cell count is usually infection somewhere in the body, but for patients with autoinflammation can indicate active disease without infection. With white blood cell count there is also a differential count performed. This looks at the specific types of white blood cells, such as neutrophils and lymphocytes, monocytes, eosinophils, and basophils.

Neutrophils are a type of white blood cell responsible for immune system response to infections or antigens (i.e. any kind of invader). Neutrophil levels may rise as a response to infection or inflammation, such as from active autoinflammatory disease, injury, other types of inflammatory processes, heart disease, or cancer. Neutrophil levels may be lower than average as a side effect of medication, or as part of a disorder's process.

Platelets are important in the blood clotting process, they are made up of fragments of a cell type called a megakaryocyte, found in the bone marrow.
Low platelet count can be caused by several things including viral infection and due to the presence of immunological disease.
High platelet count can indicate inflammation or infection.

Liver function Test (LFT's)

Alanine Transaminase (ALT) is a test that measures the levels of an enzyme found mostly in the liver. This enzyme, ALT, can also be found albeit in much lower amounts in a person's kidneys, heart and muscles. Elevated ALT levels may indicate liver damage or liver disease, but ALT may also be elevated when a person has inflammation in the bile ducts, from fatty liver, or a viral liver infection (hepatitis). It is important to note that some medication may also cause an ALT elevation; your doctor will be able to determine whether this is the case and whether any medication requires adjustment as a result. An ALT test is part of more wider liver function tests, several different blood tests that measure the levels of different proteins and enzymes that, when looked at together, can provide doctors with a comprehensive idea of how well the liver is functioning.

Alkaline Phosphatase (ALP) is a test that measures the levels of an enzyme found mostly in the liver. This enzyme, ALP, is found in much lower amounts in the bones as well. Elevated levels of ALP usually indicate liver disease, inflammation of the bile ducts or certain disease processes of the bones. It is important to note that some medication may also cause an ALP elevation; your doctor will be able to determine whether this is the case and whether any medication requires adjustment as a result. An ALP test is part of more wider liver function tests, several different blood tests that measure the levels of different proteins and enzymes that, when looked at together, can provide doctors with a comprehensive idea of how well the liver is functioning. Very high levels of ALP might indicate vitamin D deficiency. ALP levels tend to be higher in healthy growing children than in adults.

Kidneys

Urea and electrolytes (U&E)

This checks for chemicals, called electrolytes, in the blood, such as sodium (salt), potassium and magnesium. If the levels are too high or too low, this can cause abnormal heart rhythms, so it is important to adjust them if the levels aren't right.

The body makes a protein called urea which is normally broken down in the kidneys. If there is a higher level in the blood than normal, this could indicate problems with kidney function, which can affect your heart too.

Some heart medications, such as ACE inhibitors, can affect your kidneys, so this test will help your doctor decide if it is safe or appropriate to increase your dose, depending on your condition and the results of the test. *[Different types of blood tests - BHF]*

Serum Creatinine is a waste product produced by the body and filtered through the kidneys. Measuring a person's serum creatinine level can show how well their kidneys are functioning.

Blood Pressure

A Blood Pressure or BP Test, whilst strictly speaking not a blood test (i.e. no needle required) may be done as an indicator of your cardiovascular health. The result will be interpreted in conjunction with blood and urine test results, any relevant family history as well as diet. High blood pressure is common in the population ("essential hypertension"), but sometimes can be caused by medicines (e.g. steroids), or damage to the kidneys from diseases including some autoinflammatory diseases

Useful Information

GENOMICS ENGLAND - GENETIC PANELS

- **Primary immunodeficiency or monogenic inflammatory bowel disease (Version: 4.03.02.1)** [321 Gene Panel]

- **Autoinflammatory disorders (Version: 1.0)** [24 Gene Panel]

[Information correct as of April 2023]

Genetic Testing

Autoinflammatory conditions often have many overlapping symptoms, which can make diagnosis difficult. There are several gene panel tests that can be performed by a blood test. The genetic tests will look to identify genetic mutations present in an individual's DNA. Some autoinflammatory diseases have specific genetic mutations associated with them, therefore the results can help the doctors differentiate which disease a patient may have.

SOME MEDICATIONS

- Colchicine
- Prednisolone (Steroids)
- Canakinumab / Ilaris
- Anakinra / Kineret
- Tocilizumab / RoActemra
- Adalimumab/ Amgevita / Imraldi / Idacio / Yuflyma / Humira
- Entanercept / Enbrel / Benpali

Mental health

RARE MINDS

SPECIALIST RARE DISEASE COUNSELLORS
Patients and family members often tell us how helpful it is to speak to a counsellor who already understand the complexity and impact of living with a rare disease.

- ALLOCATED LEAD COUNSELLOR
- FULLY MANAGED SERVICE
- TELEPHONE & VIDEO SESSIONS
- PRIVATE THERAPY SERVICE
- COUPLES COUNSELLING
- OUTREACH AT CLINICS OR EXTERNAL SERVICES

MIND

"We provide advice and support to empower anyone experiencing a mental health problem. We campaign to improve services, raise awareness and promote understanding."

SAMARITANS

"If you need someone to talk to, we listen. We won't judge or tell you what to do"

YOUNG MINDS

"Whether you want to understand more about how you're feeling and find ways to feel better, or you want to support someone who's struggling, we can help."

Support for carers

CARERS UK

"No matter what you're experiencing as a carer, we're here to help with information, advice and support for your wellbeing."

CARERS TRUST

"Carers Trust works to transform the lives of unpaid carers. It partners with its network of local carer organisations to provide funding and support, deliver innovative and evidence-based programmes and raise awareness and influence policy. Carers Trust's vision is that unpaid carers are heard and valued, with access to support, advice and resources to enable them to live fulfilled lives."

ACTION FOR CHILDREN

"We can't take away a parent's illness, but we can give young carers a break. Our services help young people balance caring with being a child"

Symptom Tracking - why do it?

Doctor's Appointments:

- It can be helpful to have everything written down in front of you before attending a GP or consultant appointment. Otherwise, it can be easy to forget to say something about a symptom, test, or anything else that has occurred in between appointments.
- The ability to track symptoms over time can give a better indication of the severity of your illness; it can show whether there has been an upward or downward trend in your disease activity, or it can show whether what you have experienced is a flare-up or not. This is key information to show your consultant/specialist as it can impact your treatment going forward.
- This all comes under communication between yourself and the medical professional. Symptom tracking is a key way of helping them to help you.

Communication is of huge importance during appointments, but it is apparent that many patients feel underwhelmed after appointments due to a lack of communication. The Cambridge Rare Disease Network has produced a useful online flipbook showing the boundaries to good care, and how they plan to address this through the use of **Patient Passports.** Go to the link below to find out more.

Cambridge Rare Disease Network - Patient Passports

MORNING CHECK IN

I WOKE UP FEELING

Awesome **Good** **Okay** **Not good** **Horrible**

What do you want to accomplish today?

How do you want to feel today?

Today's affirmation:

Food journal

WEEK: _____

Breakfast _____
Lunch _____
Dinner _____
Snacks _____
Rate your day ○○○○○

Breakfast _____
Lunch _____
Dinner _____
Snacks _____
Rate your day ○○○○○

Breakfast _____
Lunch _____
Dinner _____
Snacks _____
Rate your day ○○○○○

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Breakfast _____
Lunch _____
Dinner _____
Snacks _____
Rate your day ○○○○○

Breakfast _____
Lunch _____
Dinner _____
Snacks _____
Rate your day ○○○○○

Notes: _____

This may be useful if you think you are sensitive towards certain foods

BLOOD MONITORING CHART

Blood Test Results	January	February	March	April	May	June
ESR						
CRP						
HB						
MCV						
WBC						
Neut						
Plats						
Lymph'						
ALT						
ALP						
Serum Creatinine						
Cholesterol						
SAA						

Blood Test Results	July	August	September	October	November	December
ESR						
CRP						
HB						
MCV						
WBC						
Neut						
Plats						
Lymph'						
ALT						
ALP						
Serum Creatinine						
Cholesterol						
SAA						

SYMPTOM DIARY

Days	Abdominal Pain	Nausea/ Vomiting	Diarrhoea	Headaches	Chest Pain	Painful glands/ Lymph nodes	Aching in limbs or joints	Swollen or red joints	Eyes manifestations	Skin Rash	Ulcers	Pain relief taken
Scored As:	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 to 3	0 or 1
1												
2												
3												
4												
5												
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29												
30												
31												

MONTH:

FEVER DIARY

Days	Morning	Inbetween	Lunchtime	Inbetween	Evening	Inbetween	Bedtime	Inbetween
Degrees Celcius								
1								
2								
3								
4								
5								
6								
7								
8								
9								
10								
11								
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31								

MONTH:

Get In Touch

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