

# The journey of a patient with a Rare Immunological Disorder

A patient journey represents the entire sequence of events that a patient experiences within a given healthcare system or across providers, from scheduling an appointment for a regular checkup to receiving treatment for an illness or injury. A patient journey is an ongoing process that incorporates all parts of the healthcare ecosystem, from hospitals to physicians, specialty care, and outpatient therapy.



Symptoms

Diagnostic



Follow-up & Aging



Treatment

Autoimmune diseases (AI)



Autoinflammatory disorders (AID)



Paediatric Rheumatic diseases (PR)



Primary Immunodeficiencies (PID)



Glossary



# Autoimmune diseases (AI)

● Autoimmune diseases (AI)

● Autoinflammatory disorders (AID)

● Paediatric Rheumatic diseases (PR)

● Primary Immunodeficiencies (PID)

**Small vessel vasculitis**



# The journey of a patient with small vessel vasculitis



**Meet Mary** she lives with one of the small vessel vasculitis conditions. Early diagnosis and appropriate timely access to treatment is essential to avoid irreversible organ damage. There are several different types of small vessel vasculitis, those which have anti-neutrophil cytoplasm antibodies – ANCA associated vasculitis (Granulomatosis with Polyangiitis, (GPA), Eosinophilic granulomatosis with Polyangiitis, (EGPA), Microscopic Polyangiitis, (MPA)) and non ANCA associated vasculitis (Henoch Schönlein Purpura/ IgA and Cryoglobulinemic Vasculitis). The main organs affected are kidneys, lungs and sinuses, but as these small vessel vasculides are systemic, there can be involvement of any organ.



Symptoms



Diagnostic



Treatment



Follow-up & aging



Resources



# Symptoms description



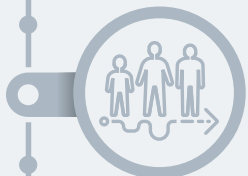
Symptoms



DIAGNOSTIC



TREATMENT



FOLLOW-UP & AGING

## ENT

- Sinusitis
- Nose bleeds
- Crusting
- Repeated infections
- Loss of hearing
- Hoarseness
- Saddle Nose

## ORAL HEALTH

- Bleeding gums
- Ulcers

## CARDIOLOGY

- Arrhythmia
- Spasm of blood vessels
- Heart failure
- Heart Oedema

## GASTRO-ENTEROLOGY

- Diarrhoea
- Nausea
- Bleeding
- Pain
- Perforated bowel (rare)

## NEPHROLOGY

- Haematuria
- Frequent kidney infections
- Urine foaming
- Increased Blood pressure

## NEUROLOGY

- Loss or alteration of sensation
- Difficulty with co ordination
- Difficulty moving around

## OPHTHALMOLOGY

- Bloodshot eyes
- Sensitivity to light
- Inflammation of eye and eye structures
- Conjunctivitis
- Changes to vision

## RESPIRATORY

- Wheeze
- Difficulty breathing
- Cough
- Coughing up blood
- Asthma
- Repeated chest infections

## DERMATOLOGY

- Rash
- Ulcers
- Necrosis

## NUTRITION

- Loss of appetite
- Loss of weight

## INFECTIONS

- Swollen glands
- Feeling unwell
- Fever
- Recurrent infections not responding to antibiotic treatment

## RHEUMATOLOGY

- Joint pain
- Joint swelling
- Fatigue
- Muscle pain



## REQUIRED PRACTITIONERS

General /Healthcare Practitioner

Nurses

A range of organ specialists linked to main presenting symptoms

Emergency ward/ A&E/ ICU

Medical assessment unit

Physiotherapy

Dentist

Optician

Alternative medicines

# Symptoms Challenges



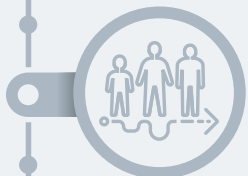
## Symptoms



## DIAGNOSTIC



## TREATMENT



## FOLLOW-UP & AGING



### HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

These are rare conditions so there is poor awareness of Vasculitis within the healthcare system especially primary care.

Symptoms come and go and a lot of symptoms are dismissed – regarded as psychosomatic.

Vasculitis mimics other illnesses.



### ACCESS TO EARLY, ACCURATE DIAGNOSIS

Diagnosis odyssey of rare condition due to poor awareness (as above)

Risk of permanent organ damage without timely intervention.

No definitive diagnostic tool.



### COMMUNICATION AND COORDINATION

These are systemic conditions resulting in many HCPs being involved in care. These may not always be aware of each other's intervention.



### HEALTH IMPACT

A wide range of very diverse physical symptoms.

Wide range of variation between individuals.

Symptoms can come and go.

Appears in childhood, adolescence as well as adulthood (peak incidence is age 50–60 years)



### PSYCHOLOGICAL & SOCIAL IMPACT

Impact on both patient and carer

Anxiety, Depression  
Isolation and loss of self-confidence as symptoms increase and interfere with independence.

Financial challenges

Absences from work or from school.

Costly health interventions



### QUALITY OF LIFE

Uncertainty re what is happening and what the future holds

Reduced independence

# Symptoms Needs



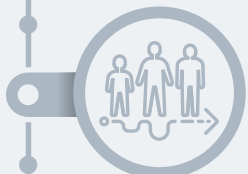
## Symptoms



## DIAGNOSTIC



## TREATMENT



## FOLLOW-UP & AGING



### HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Increase awareness of small vessel vasculitis amongst the healthcare professionals.

Embedding Vasculitis in HCP and Medical student education modules.

Increase awareness and encourage attendance of existing specialist Vasculitis courses.



### COMMUNICATION AND COORDINATION

Improve onward referral to other disciplines and improve communication between the same.

Collaboration between healthcare professionals involved in care to achieve earlier diagnosis and access to treatment.

Encourage holistic view of the patient, not the individual symptoms.



### DIGITAL HEALTH

All involved healthcare professionals to have access to health care records to ensure continuity of care.



### SUPPORT & ADVOCACY

Support patient and family at this difficult time as they await a diagnosis .

Refer to local agencies who may be able to help with support and financial challenges.

Link with education service to arrange alternative solutions to overcome absence from school.

# Diagnostic description



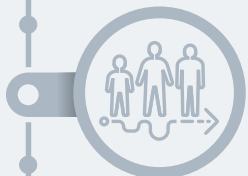
SYMPTOMS



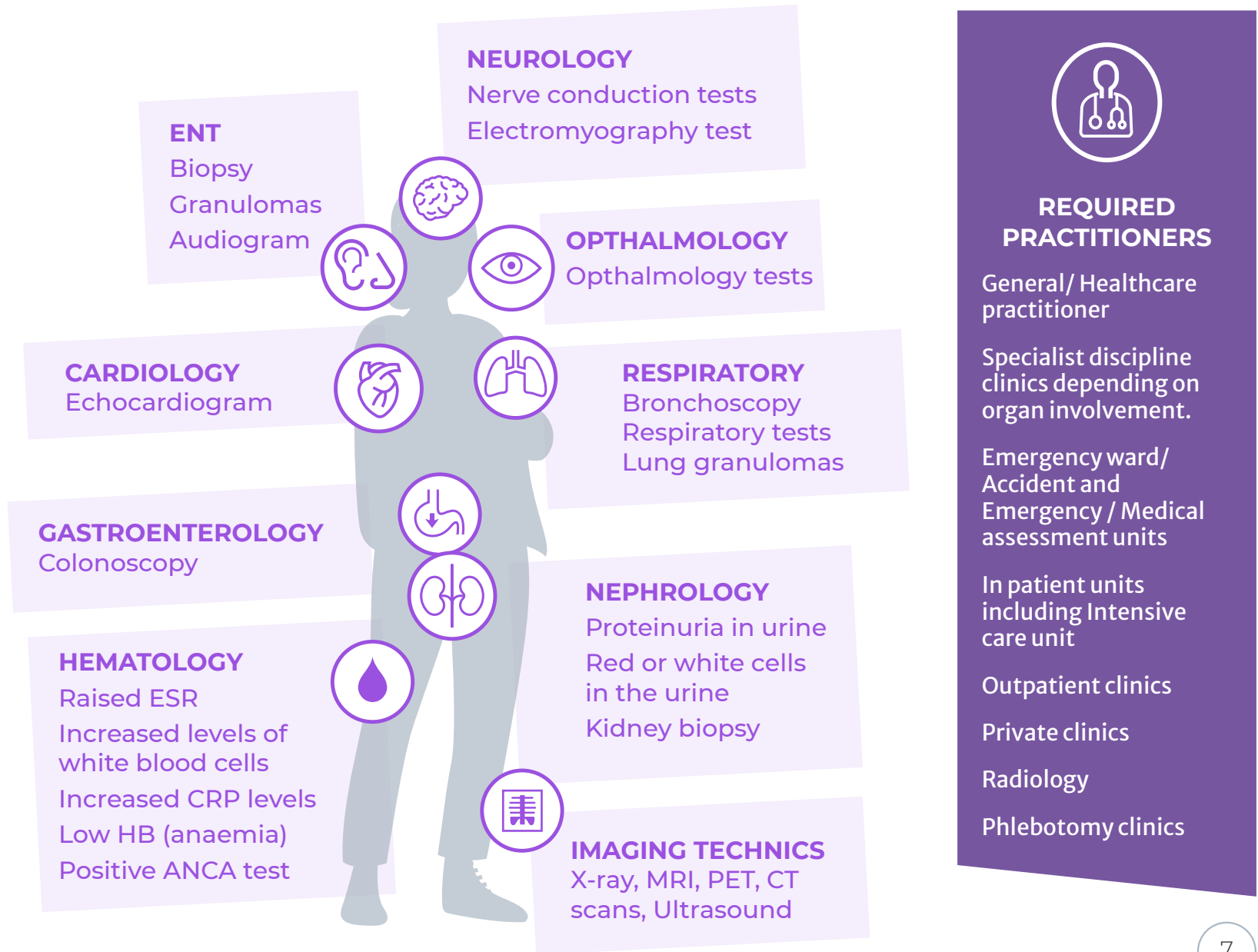
Diagnostic



TREATMENT



FOLLOW-UP & AGING



Clinical presentation of all presenting symptoms and acknowledgement of the same.

# Diagnostic Challenges



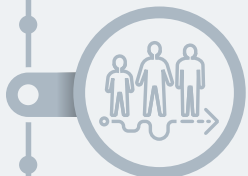
SYMPTOMS



## Diagnostic



TREATMENT



FOLLOW-UP & AGING



### HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Lack of awareness leads to delayed access to appropriate tests, results and Diagnosis.

Unfortunately, small vessel vasculitis can mimic some illnesses which leads to misdiagnosis.



### HEALTH IMPACT

A wide range of very diverse physical symptoms.

Wide range of variation between individuals.

Symptoms can come and go.

Appears in childhood, adolescence as well as adulthood (peak incidence is age 50–60 years).



### ACCESS TO EARLY, ACCURATE DIAGNOSIS

Delayed diagnosis can lead to permanent organ damage.

There is no definitive diagnostic test and so we rely on combining results from various tests to get an accurate picture.

Delayed diagnosis can lead to chronic repeat infections which reactivate the disease causing more damage.



### COMMUNICATION AND COORDINATION

Many specialists are involved and need to have clear communication (digital health systems) in place to ensure holistic picture of patient and efficient use of available resources.



### PSYCHOLOGICAL AND SOCIAL IMPACT

Impact on both patient and carer

Anxiety, Depression  
Isolation and loss of self-confidence as symptoms increase and interfere with independence.

Financial challenges.

Absences from work or from school.

Costly health interventions.



### QUALITY OF LIFE

Uncertainty re what is happening and what the future holds.

Reduced independence



# Diagnostic Needs



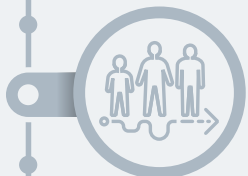
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING



## HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Increase awareness of signs of small vessel vasculitis amongst the healthcare professionals.

Embedding Vasculitis in HCP and Medical student education modules.

Increase awareness and encourage attendance of existing specialist Vasculitis courses.



## RESEARCH

National and International cooperation – establishment of networks.

Data Sharing and interoperability.

Further research in genomics and biomarkers to establish clear diagnosis tools.

Registries

Reevaluation of diagnosis in time given the progress in the field including genomics and biomarkers.



## SUPPORT & ADVOCACY

Patient needs to feel able to advocate for themselves to ensure getting the right diagnosis.

Signpost to relevant local services to help with all practicalities of diagnosis, physical, psychological and social.- including advise re work, education, or planning for early retirement.

Signpost to local support group so link with others living with the condition.

Signpost to any benefit system that may assist with any associated financial costs of diagnosis.

Psychological support via counselor, psychologist, and/ or self-help groups.

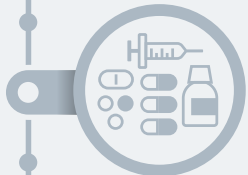
# Diagnostic Needs



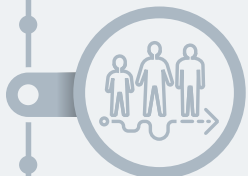
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING



## COMMUNICATION & COORDINATION

Collaboration between clinics.

.....  
 Clear explanations of what is happening to patient and carer.



## QUALITY OF LIFE

Access to support services as outlined above.

.....  
 Reassurance that one can recover a good quality of life with treatment.

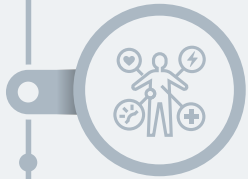


## ACCESS TO EARLY, ACCURATE DIAGNOSIS

Access to tests and prompt results  
 Timely intervention.

.....  
 Access to specialist Clinicians/ specialist small vessel vasculitis clinic.

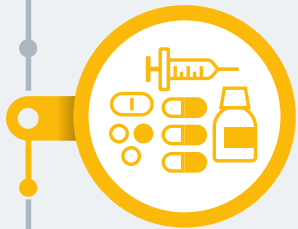
# Treatment description



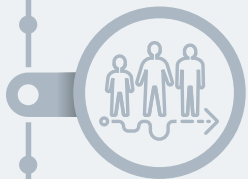
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING

## IMMUNOSUPPRESSION

Immunosuppression by Steroids, Chemotherapy and Biological drugs DMARDS Disease modifying anti-rheumatic drugs



## PHYSIOTHERAPY

## OCCUPATIONAL THERAPY

## ALTERNATIVE THERAPIES

## SENSORY SUPPORT APPLIANCES

## ANTIBIOTICS

either prophylactic or to treat infection



## PAIN, FATIGUE AND BREATHLESSNESS MANAGEMENT

## KIDNEY DIALYSIS AND TRANSPLANT



## PSYCHOLOGICAL SUPPORT



## SURGICAL INTERVENTION



## TREATMENT OF SIDE EFFECTS AND PROPHYLACTIC MEASURES

to prevent escalation of same : osteoporosis, gastric ulcers,

## VACCINATIONS

Flu, Pneumonia, Covid-19



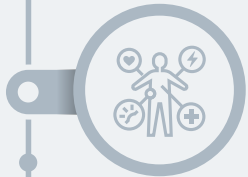
## PODIATRY



## REQUIRED PRACTITIONERS

- General/ Healthcare practitioner
- Specialist discipline clinics depending on organ involvement
- In patient clinics
- Outpatient clinics
- Phlebotomy clinics
- Family planning clinics
- Private clinics
- Specialised nurses
- Community nurses
- Hospital and community pharmacists
- Physiotherapist
- Occupational Therapist
- Podiatrist
- Dentist
- Dietitian
- Social worker
- Psychological support
- Alternative therapies

# Treatment Challenges



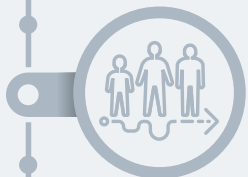
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING



## HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Lack of awareness leads to delays in treatment and possible organ damage.



## ACCESS

Sometimes special permission/approval/funding is required to access novel treatments

May need travel to clinics to have treatments and these may be some distance from home.



## COMMUNICATION AND COORDINATION

Because so many different disciplines are involved, we need to have clear communication between the all disciplines involved.



## CHRONICITY OF TREATMENT

Immunosuppression leads to increased risk of infection and so restrict quality of life (education, employment, social situations)

Use of chemotherapy has implications on fertility and can induce early menopause.

Many treatments have side effects which need further monitoring /treating.

Long term use of steroids leads to development of cushingoid appearance.

Long term use of treatments leads to development of co morbidities i.e., osteoporosis, diabetes, glaucoma, increased risk of developing cancer.



## QUALITY OF LIFE

Determined by organ involvement, independence, and ability to resume previous roles.



## PATIENT EMPOWERMENT

Patient and their families are adjusting to a new life with small vessel vasculitis, how it impacts them, how the treatment impacts them and the changes that are needed in their lives. This is a very isolating and vulnerable time.



## PSYCHOLOGICAL AND SOCIAL IMPACT

Lack of psychological support leads to anxiety re life changes, loss of independence, confidence, and self-esteem, stress related flare ups, and poor social interaction.

# Treatment Needs



SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING



## ACCESS

Clear procedures in place to enable appropriate and timely access to current treatments.



## HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Increase awareness of treatment of small vessel vasculitis amongst the healthcare professionals.

Embedding Vasculitis in HCP and Medical student education modules.

Increase awareness and encourage attendance of existing specialist Vasculitis courses.



## COMMUNICATION AND COORDINATION

Clear established communication between all disciplines involved (i.e. electronic healthcare records).



## CHRONICITY OF TREATMENT

Clear guidance on how to avoid exposure to infections.

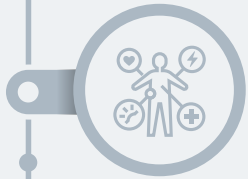
Appropriate prophylactic measures in place to avoid development of co morbidities.

Advise re fertility, and options available re egg/sperm storage.

Advise on managing the menopause for women exposed to chemotherapy.

Treatment /management of side effects.

# Treatment Needs



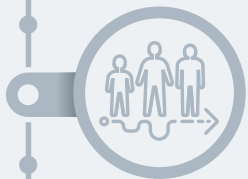
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP  
& AGING



## SUPPORT AND ADVOCACY

Involvement in support group and/or self-help groups to empower the individual and their family in managing their condition and the related issues.



## PATIENT ENGAGEMENT

Need to have clear communication with the patient and their carer to plan, explain and execute the appropriate treatment plan for the individual.

Information leaflets are usually available on the specific conditions and drugs, but these need reinforced by one-to-one discussion to explain importance of drug / treatment regime, expected side effects, and precautions to be aware of.

The care team should be aware of the implications of small vessel vasculitis and its treatment on the life of the individual and their family – to this end the treatment plan should be a collaboration between the individual (and their family in the case of a child) and their clinicians.



## RESEARCH

Clinical trial to test new drugs/ interventions and regimes of treatment to improve quality of life and manage small vessel vasculitis.

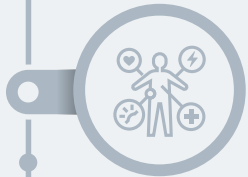
National and International cooperation – establishment of networks

Data Sharing and interoperability.

Further research to establish tools to identify flares, how to avoid flares and new treatments. – genomics, biomarkers

Registries to determine incidence, disease progression and effective treatments.

# Follow up & Ageing description



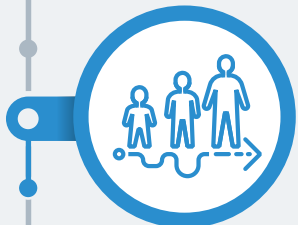
SYMPTOMS



DIAGNOSTIC



TREATMENT



Follow-up & Ageing

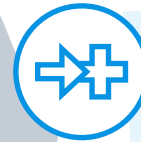
**INDIVIDUAL AWARE**  
of symptoms of relapse  
and what to do when  
feeling unwell



**SHARED CARE**  
between specialist clinics  
and general healthcare  
practitioner



**REFERRAL TO  
FURTHER CLINICS  
AS INDICATED**



**DEVELOPMENT  
OF CO  
MORBIDITIES**  
and address issues



**REGULAR REVIEWS**  
by general healthcare  
practitioner, the named  
specialist consultant, and  
their team at all discipline.  
Clinics involved in care



## REQUIRED PRACTITIONERS

General Healthcare practitioner.

In patient clinics

Out patient clinics

Specialist nurses

Community nursing

Pharmacist

Emergency ward

Healthcare professionals involved in care.

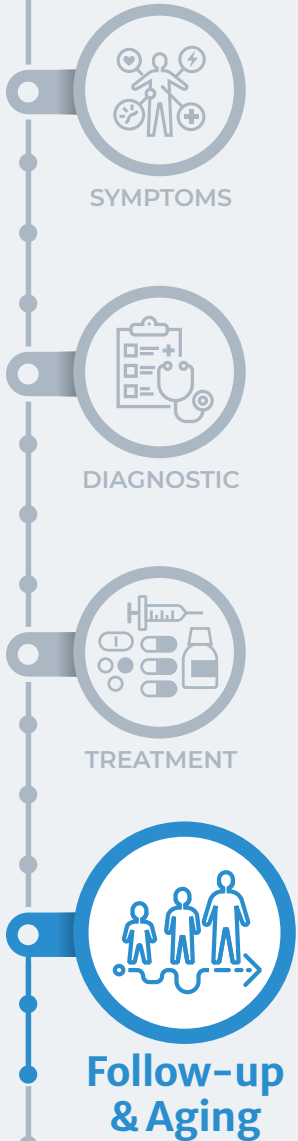
Social worker

Transition clinics

Gerontologist

# Follow up & Ageing Challenges

The journey of a patient with small vessel vasculitis



## MEDICAL FOLLOW UP

Regular monitoring, access to test results and ability to quickly report symptoms to medical team to avoid unnecessary worry.



## SUPPORT AND ADVOCACY

Access financial help/benefits and help to adjust to new life within the family.

Housing/school/workplace adaptations.

Physical/financial and psychological support systems in place



## HCP FOLLOW UP

Multi system illness so need access to many different disciplines.

Access to HCPs as needed.

Access to Transition clinic for children.

Access to gerontology services in old age.



## PATIENT EDUCATION

Awareness of what to do if symptoms change.

Awareness of how to manage condition by medication, lifestyle, and self-help.



## CHRONICITY OF TREATMENT

Immunosuppression leads to increased risk of infection and so restrict quality of life (education, employment, social situations).

Use of chemotherapy has implications on fertility and can induce early menopause.

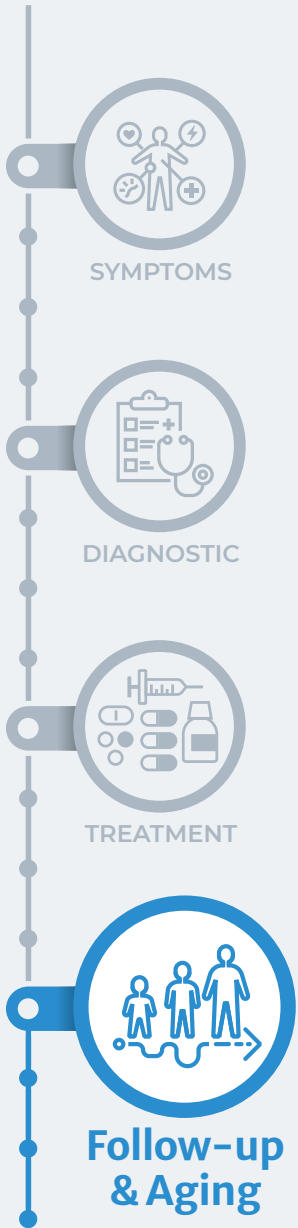
Many treatments have side effects which need further monitoring /treating.

Long term use of steroids leads to development of cushingoid appearance.

Long term use of treatments leads to development of co morbidities i.e., osteoporosis, diabetes, glaucoma, increased risk of developing cancer.



# Follow up & Ageing Needs



## MEDICAL FOLLOW UP

Care needs to be coordinated by one clinician – rheumatologist, nephrologist or immunologist, and the individual patient. .



## MEDICAL AWARENESS AND EDUCATION

Need to be aware of the changes that accompany aging and how to address these.

Plan for smooth transition of care from childhood services to adult and from adult to gerontology.



## SUPPORT AND ADVOCACY

Support individual, carer and families in adjusting.



## HCP FOLLOW UP

Good communication to exist between all involved in patient care.



## PATIENT EDUCATION

Individuals and their family need to have more information on medications, treatments and living with small vessel vasculitis long term.



## DIGITAL HEALTH

IT tools like dedicated smartphone apps allowing for real time reporting about disease symptoms and treatment complications to medical team.

Virtual clinics when condition is stable to avoid having to travel to busy clinics.



## RESEARCH

Need registries to gather information on effective management of small vessel vasculitis, effective treatments, disease progression and geographical incidence.

Clinical trials.

Exploration of genomics and biomarkers in small vessel vasculitis.

**Healthcare professionals' awareness and education ERN RITA – CPMS** service – virtual service providing advice from specialist clinicians in response to query from local individual HCP.

**EULAR** training courses Specialising in Vasculitis.

**UKIVAS** training courses specialising in Vasculitis.

**EUVAS** – guidelines and information on Vasculitis.

**Vasculitis Foundation/Vasculitis UK/ local vasculitis patient organisations**– information on Vasculitis.

**Support and Advocacy Vasculitis International** – list of patient support groups and vasculitis clinics throughout Europe

**Vasculitis International** – list of patient support groups and vasculitis clinics throughout Europe.

**Local support services** re financial hardship, employment issues, improved independence, and mental well-being.

Patient Education Above mentioned support groups

**Myancavasculitis.com** –website created by patients, pharma and clinicians giving useful information on AAV.

**Access to early, accurate diagnosis Birmingham Vasculitis Activity Score**– <https://vasculitis.org/disease-scoring/>

**Vasculitis Damage Index** – <https://vasculitis.org/disease-scoring/>

**Chapel Hill Classification of Vasculitis**



# Primary Immunodeficiencies (PIDs)

Autoimmune diseases (AI)

Autoinflammatory disorders (AID)

Paediatric Rheumatic diseases (PR)

**Primary Immunodeficiencies (PID)**

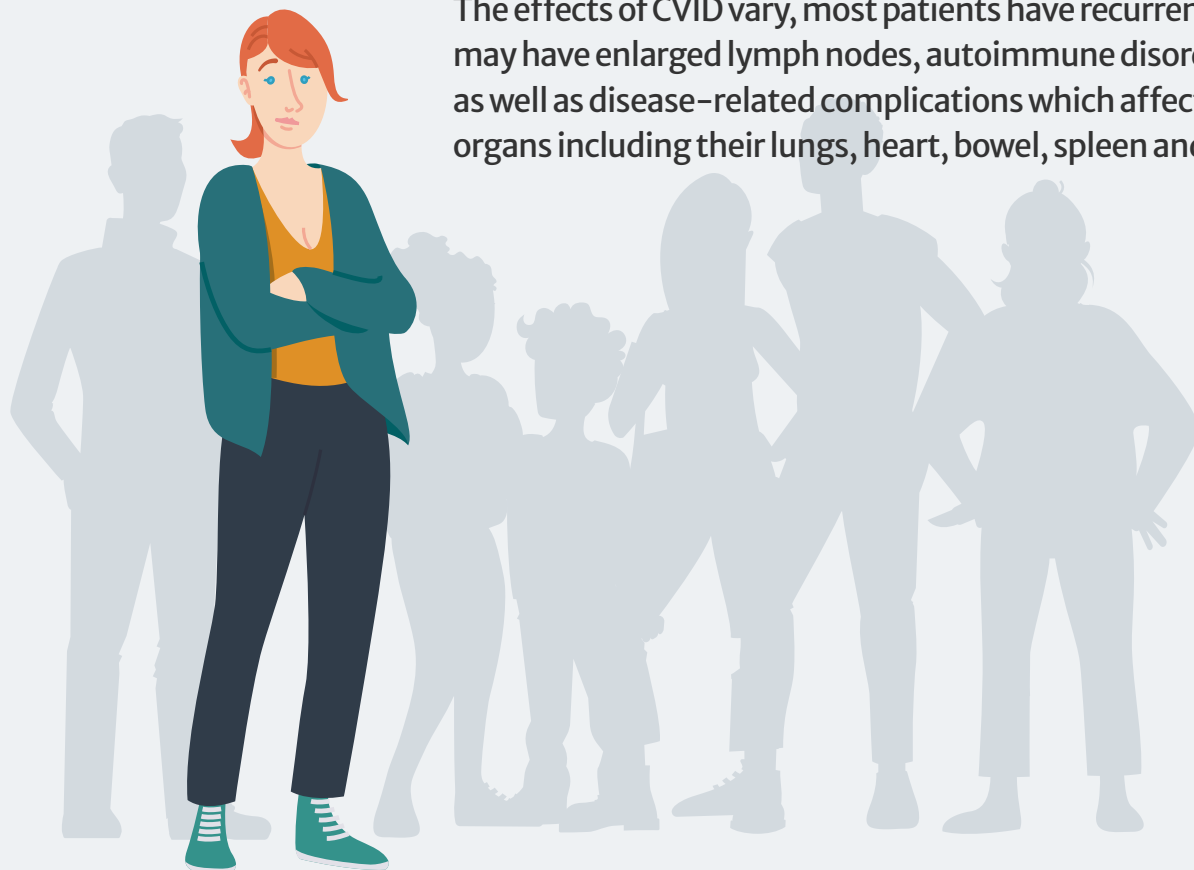
## Common Variable Immune Deficiency (CVID)



# The journey of a patient with CVID

**Meet Helen** She is a person with **Common Variable Immune Deficiency (CVID)**, an heterogeneous group of diseases characterised by a significant failure to produce specific antibodies and susceptibility to bacterial infections.

The effects of CVID vary, most patients have recurrent infections, and some may have enlarged lymph nodes, autoimmune disorders as well as disease-related complications which affect their organs including their lungs, heart, bowel, spleen and liver.



Symptoms



Diagnostic



Treatment



Follow-up & aging



Resources



# Symptoms description



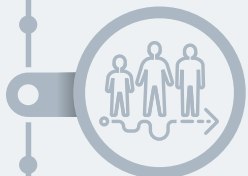
## Symptoms



## DIAGNOSTIC



## TREATMENT



## FOLLOW-UP & AGING

### GENERAL

Fever  
Fatigue

### ENT

Sinusitis  
Otitis  
Tinnitus  
Loss of hearing due to infections  
Dizziness in case of otitis  
Allergy

### NUTRITION

Eating disorders  
Malabsorption  
Denutrition  
Obesity

### GASTROENTEROLOGY

Gastro enteritis  
Splenomegaly  
Hepatomegaly  
Loss of appetite,  
Nausea, vomiting  
Weight loss  
Elevated liver enzymes  
Abdominal pain/discomfort  
Granulomas (liver, gut)  
Digestive ulcers  
Chronic/recurrent diarrhoea  
Mucus and blood in stool (infection-related or inflammatory bowel disease related)  
Infections (bacteria/viruses)

### HAEMATOLOGY

Pancytopenia (anemia, thrombocytopenia, lymphopenia)  
Lymphadenitis  
Haemolytic anemia  
Leukocytopenia  
Very low IgG level  
Very low IgM level  
Very low IgA level  
Low IgG subclasses  
Lymphomas/Leukemia  
Hepato-splenomegaly

### NEUROLOGY

Development/growth delay  
Meningitis  
Gait/motor alterations

### DERMATOLOGY

Skin infections (folliculitis, infections, delayed scarring, warts)  
Vasculitis  
Purpura/petechiae  
Muscle skin layer painful  
Chronic/recurrent eczema  
Bruising  
Silver hair, fragile hair  
Ombilical cord issues

### RESPIRATORY

Chronic cough  
Dyspnea, Shortness of breath, ...  
Sputum production  
Lung problems  
Intrapulmonary lesions (nodular lesion)  
Recurrent bronchitis and pneumonias  
Bronchiectasis  
Pneumatocele  
Interstitial lung disease  
Asthma

### RHEUMATOLOGY

Moving joint pains or inflammation  
Arthritis  
Osteopenia (in children/young)  
Osteoporosis  
Recurrent tendinitis

### INFECTIONS

Sepsis  
Long lasting/severe/repeated varicella  
Recurrent/More severe/Atypical or opportunistic infections  
Lymph nodes  
Shingles  
Repeated influenza  
Recurrent fever/inflammation  
Need of intravenous antibiotics



## REQUIRED PRACTITIONERS

- General Practitioners
- Paediatricians
- Internists
- Microbiologists
- Organ specialists:
  - Pulmonologist,
  - Digestive,
  - Dermatologist,
  - Hematologist,
  - Neurologist,
  - Rheumatologist,
  - Haematologists,
  - Immunologists,
  - Infectiologists,
  - Allergists
  - Dentists,
  - Ophthalmologists
- Emergency ward

# Symptoms Challenges



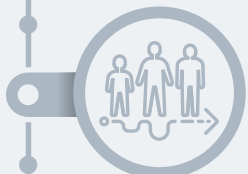
## Symptoms



### DIAGNOSTIC



### TREATMENT



### FOLLOW-UP & AGING



### HEALTH IMPACT

A wide range of very diverse symptoms:

- Varying from person to person
- Possibly changing throughout life
- Appearing in childhood as well as in adulthood

Various situations of onset:

- After repeated infections, pneumonias
- After autoimmune cytopenia
- After lymphadenopathy or splenomegaly enlargement
- After chronic diarrhoea
- After vaccine adverse reactions or infections
- After given birth
- After being in the sun
- After exercising



### ACCESS

Symptoms are addressed rather than the underlying condition that is not diagnosed



### ADDED RISKS

Risk of permanent organ damages (bronchiectasis/malignancy/death)



### HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Doctors don't know about PIDs (ie when hospitalised...)

Lack of immunologist



### PSYCHOLOGICAL & SOCIAL IMPACT

Living with a chronic invisible condition

Being regularly unwell

Being afraid of catching infections

Absences from school

Absences from work

Family burden (care-takers)

Stigma because of unusual symptoms

People being always ill, but not being believed they are ill

Financial challenges (ability to work)

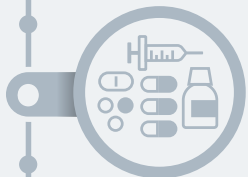
# Symptoms Needs



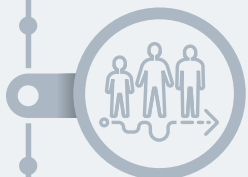
Symptoms



DIAGNOSTIC



TREATMENT



FOLLOW-UP  
& AGING



ACCESS

Early accurate diagnosis

Immunologists for children  
and adults

Expertise centres



HEALTHCARE  
PROFESSIONALS  
AWARENESS & EDUCATION

Awareness on warning signs

Medical education of general  
practitioners and other  
healthcare professionals



RESEARCH

Research on these conditions

Publications



SUPPORT &  
ADVOCACY

Patient organisations



QUALITY OF LIFE

Possibility for home schooling  
or available distance learning  
service from school

Possibility for home working

# Diagnostic description



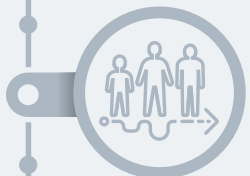
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING

## BIOLOGICAL

Decrease levels of immunoglobulins with abnormalities  
count of B cells  
Autoimmunity panels  
Microbiological tests

## IMMUNISATION

Immunisation response

## IMAGING TECHNIQS

## GENETIC TESTING

## CLINICAL

Family history  
Prenatal study  
Birth screening  
Infection history



## REQUIRED PRACTITIONERS

PID specialists  
Immunologists  
Organ specialists  
Biologists  
Psychologists  
Social workers



# Diagnostic Challenges



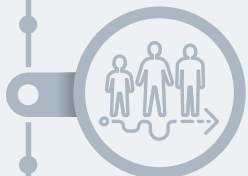
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP  
& AGING



## ACCESS

Patient odyssey before accurate diagnosis

Symptoms can appear before the age of 3, but no definitive diagnosis possible before age of 4

Misdiagnosis

Often not diagnosed until adulthood

Genetic counselling

Genetic testing



## PSYCHOLOGICAL & SOCIAL IMPACT

Difficulty for the patient/parents to accept the chronicity of the disease and of the treatment



## HEALTH IMPACT

Severe, unusual, recurrent bacterial infections

Late diagnosis can lead to permanent organ damages  
Need of ENT (Ear, Nose, Throat) surgery(ies)



## ADDED RISKS

Possible associated conditions

- Auto inflammation
- Auto immunity
- Allergy
- Malignancy

Be hospitalised for another reason than the PID in the emergency ward where doctors don't know about PIDs and may not listen to the patient /parent



## HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Many specialists, no diagnosis

Not taken seriously by the doctors even addressed to a psychiatrist

Lack of medical knowledge on PIDs

Difficult to find doctors who know about PIDs

Lack of immunologists

Lack of Adult immunologists experts in PIDs

PIDs, and especially CVIDs, are ignored by most curricula in many countries

Genetic counselling

# Diagnostic Needs



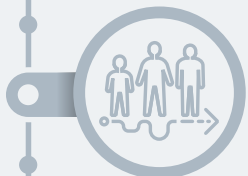
SYMPTOMS



Diagnostic



TREATMENT



FOLLOW-UP & AGING



## ACCESS

Timely and accurate diagnosis

.....  
A network of expertise centres



## MEDICAL PRACTICE

Reevaluation of diagnosis in time given the progress in the field (including the possibility of genetic testing)

.....  
When an indication for psychiatric drugs, need of psychiatry consultation



## SUPPORT & ADVOCACY

Patient advocacy organisations

.....  
Stakeholders' cooperation



## DIGITAL HEALTH

Registries

.....  
Data sharing and interoperability



## COMMUNICATION & COORDINATION

International Cooperation



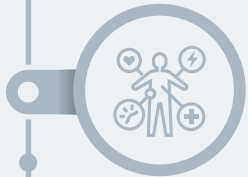
## HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Need of Immunologists for children and adults (immunology not always considered as a subspeciality)

.....  
A range of medical specialists knowing CVIDs, including biologist, geneticist, infectiologist, psychologist, ...

.....  
Immunology: More education for medical students, GPs, specialists at pre and postgraduation

# Treatment description



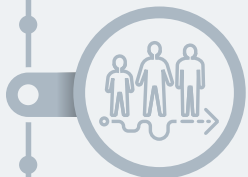
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING

IMMUNOGLOBIN REPLACEMENT THERAPY REGULAR AND LIFELONG



VACCINES (specific for pid)



ANTIBIOTICS (prophylaxis or on-demand)



SPECIFIC MEDICINES addressing specific symptoms



## REQUIRED PRACTITIONERS

PID specialists

Organ specialists knowing PIDs

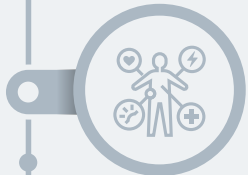
Specialised nurses

Other Healthcare professionals (physiotherapist, nutritionist, dentists...)

Psychologists / Psychiatrists

Social workers

# Treatment Challenges



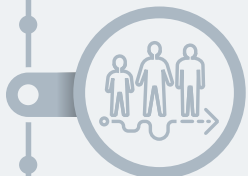
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING



## ACCESS

Supply tensions on immunoglobulins (Subcutaneous (SC) and Intravenous (IV))

Access to best tolerated immunoglobulin therapy (SC and IV)

Supply tensions/shortages on antibiotics

Access to efficient and innovative therapies (anti-infectious, targeted therapies, ...)

Access to off label immune suppressive drugs

Access to home therapy



## CHRONICITY OF TREATMENT

Treatment not well tolerated

Distance to travel to reach out to hospital for regular treatment

Distance to travel to get the medicine from hospital for home therapy



## ADDED RISK

Microbial multi resistance



## QUALITY OF LIFE

Fatigue

Time taken on family, work and social life for recurrent treatment



## PATIENT EDUCATION & EMPOWERMENT

Patient compliance to their chronic treatment

# Treatment Needs



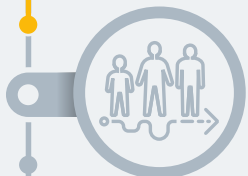
SYMPTOMS



DIAGNOSTIC



Treatment



FOLLOW-UP & AGING



## ACCESS

Continuous supply of needed medicines

Cost coverage of needed medicines

Patient's and physician's (not hospital's) choice of treatment regarding clinics and quality of life



## HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

Adapted medical devices (especially in children)

To take pain into account when administrating the treatment



## MEDICAL PRACTICE

Personalised treatment

Multidisciplinary teams

To adress side effects of treatment

Protocols to adjust therapy dosing or to withdraw anti-inflammatory therapy

Protocols on oncologic treatment in PID

Emergency protocols for PID patients



## QUALITY OF LIFE

Facilitating home therapy after patient's choice (availability, trained professionals, patients' training, ...)



## PATIENT EDUCATION & EMPOWERMENT

To understand their disease, cope with it, be compliant with treatment

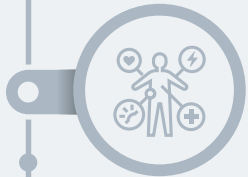


## RESEARCH

On new ways to fight off multi drug resistant bacterias

On vaccines

# Follow up & Ageing **description**



SYMPTOMS



DIAGNOSTIC



TREATMENT

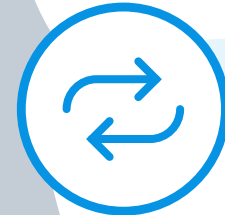


**Follow-up & Aging**

**FOLLOW-UP OF SPECIFIC ORGANS**  
(ie lung, liver...)



**RE-EVALUATE TREATMENT REGULARLY**



**TRANSITIONING**



**REGULAR LIFELONG VISITS WITH A PID SPECIALIST**  
(vary from every 3 months to every 2 years or more)

**ADDED CO-MORBIDITIES LINKED TO AGE**



**REQUIRED PRACTITIONERS**

PID specialists

Organ specialists knowing PIDs

Specialised nurses

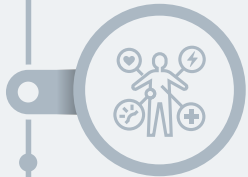
Emergency ward

Other Healthcare professionals (physiotherapists, nutritionists, ...)

Gerontologists

Psychologists

Social Workers



SYMPTOMS



DIAGNOSTIC



TREATMENT



Follow-up & Aging



## ACCESS

Access to treatment

Distance to travel to reach out to specialists



## MEDICAL PRACTICE

Onset of new symptoms and biomarkers

Lymphomas and other malignancies

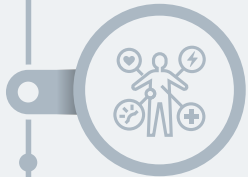
Processes and organisation for transitioning from paediatric to adult ward

Management of co-morbidities

Immunologists for adults or internists knowing PIDs/CVIDs

Identifying other specialists knowing PIDs

# Follow up & Ageing Needs



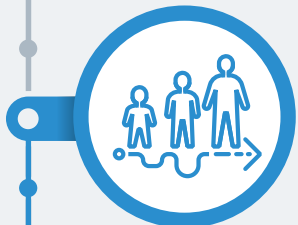
SYMPTOMS



DIAGNOSTIC



TREATMENT



Follow-up & Aging



## MEDICAL PRACTICE

A range of medical specialists knowing COVIDs

.....

Transitioning programmes for adolescent and aging persons

.....

Follow-up protocols



## DIGITAL HEALTH

Telemedicine



## COMMUNICATION AND COORDINATION

Communication between local hospitals/GPs and reference centres to ensure an optimal environment for the management of the condition and emergencies

.....

Multidisciplinary team taking a holistic approach



### International Patient Organisations

The following organisations provide a large amount of information and support material addressing their different audiences.

They also give insights on other organisations existing at national level.

#### ERN-RITA (<https://ern-rita.org/>)

The European Reference Network that aims at improving the care of patients with Rare Immunological Disorders. One stream of ERN-RITA addresses Primary Immunodeficiencies. RITA brings together leading European centres with expertise in diagnosis and treatment of rare immunological disorders. These rather potentially life-threatening conditions require multidisciplinary care using complex diagnostic evaluation and highly specialised therapies for all four streams.

#### IPOPI (<https://ipopi.org/>)

The International Patient Organisation for Primary Immunodeficiencies is the Association of national primary immunodeficiency (PID) patient organisations and the leading advocate for PID patients worldwide working in collaboration with patients, doctors, politicians, regulators, pharmaceutical industry and other relevant stakeholders.

#### ESID (<https://esid.org/>)

The European Society for immunodeficiencies is a medical society which is striving to improve the knowledge in the field of Primary Immunodeficiency (PID) by encouraging research, developing educational programs and fostering cooperation among all those involved in the diagnosis, treatment and management of these diseases.

### Tools

- PID Life Index (<https://pidlifeindex.ipopi.org/#/en/principles/world-map>)
- The PID Life Index is an interactive tool built on 6 key principles of care that measures the status of the PID healthcare environment in a country.
- Clinical guidelines
- Common Variable Immunodeficiency diagnosis criteria <https://esid.org/Education/Common-Variable-Immunodeficiency-CVI-diagnosis-criteria>

# Glossary

In process