

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



Primary Immunodeficiencies (PIDs)

● Autoimmune diseases (AI)

● Autoinflammatory disorders (AID)

● Paediatric Rheumatic diseases (PR)

● **Primary Immunodeficiencies (PID)**

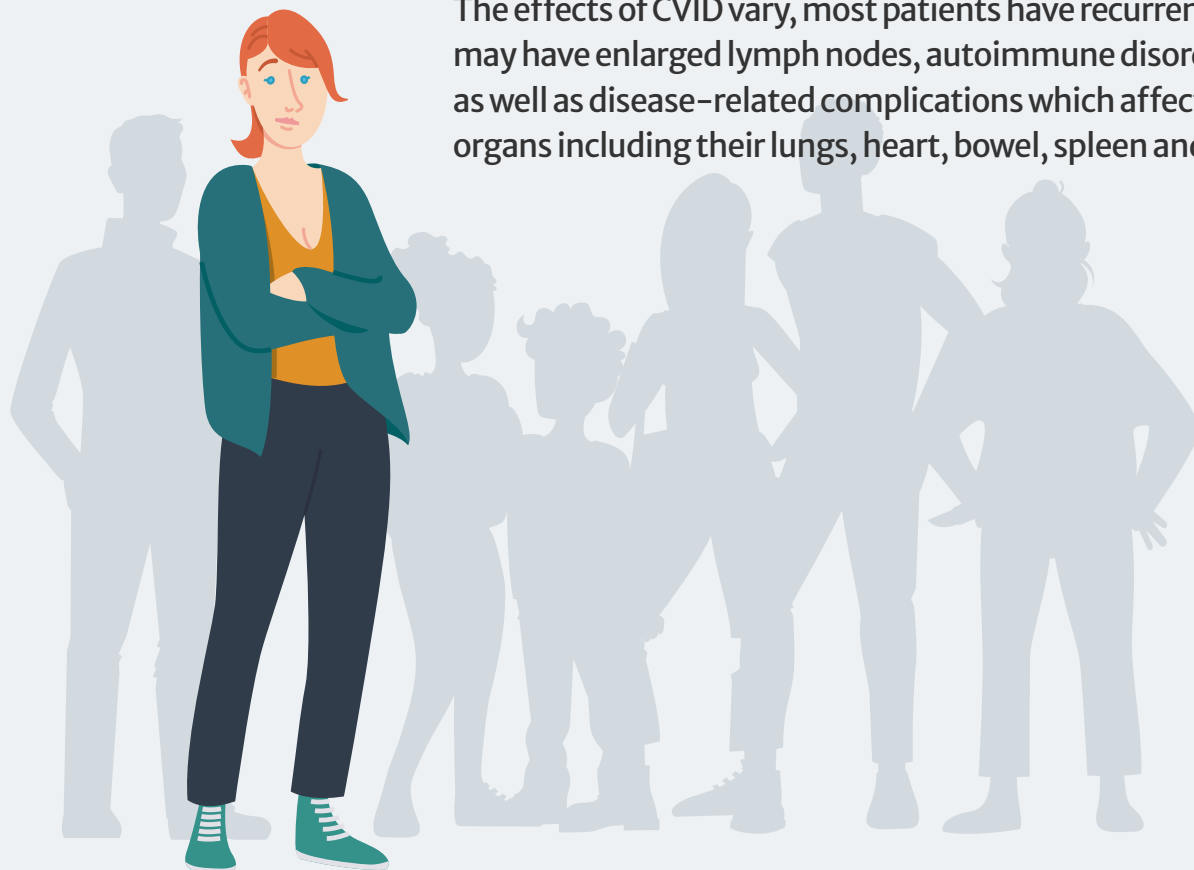
Common Variable Immune Deficiency (CVID)



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



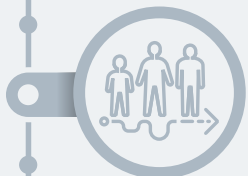
Symptoms



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



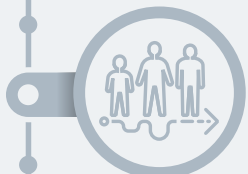
Symptoms



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)

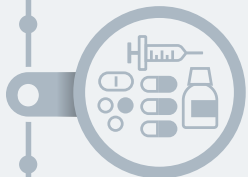
Symptoms Needs



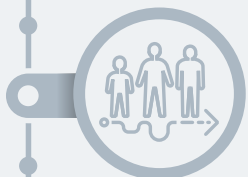
Symptoms



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

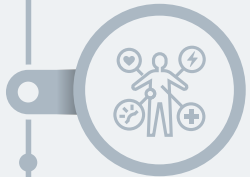


QUALITY OF LIFE

Possibility for home schooling
or available distance learning
service from school

Possibility for home working

Diagnostic description



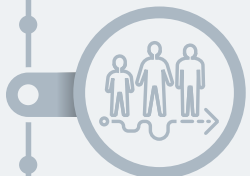
SYMPTOMS



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

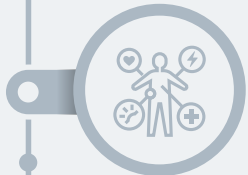
Family history
Prenatal study
Birth screening
Infection history



REQUIRED PRACTITIONERS

PID specialists
Immunologists
Organ specialists
Biologists
Psychologists
Social workers

Diagnostic Challenges



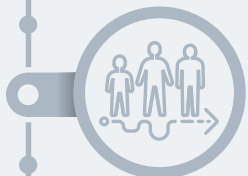
SYMPTOMS



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



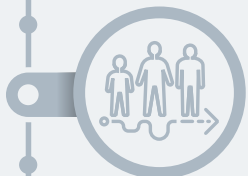
SYMPTOMS



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



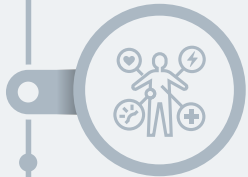
HEALTHCARE PROFESSIONALS AWARENESS & EDUCATION

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

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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



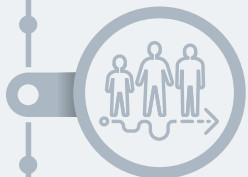
SYMPTOMS



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



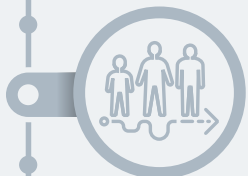
SYMPTOMS



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

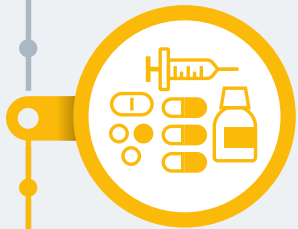
Treatment Needs



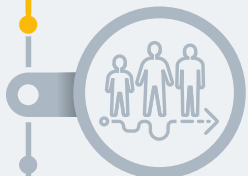
SYMPTOMS



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

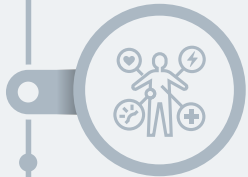


RESEARCH

On new ways to fight off multi drug resistant bacterias

On vaccines

Follow up & Ageing **description**



SYMPTOMS



DIAGNOSTIC



TREATMENT



Follow-up & Aging

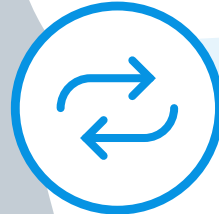
FOLLOW-UP OF SPECIFIC ORGANS
(ie lung, liver...)



RE-EVALUATE TREATMENT REGULARLY




TRANSITIONING

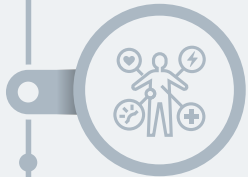


REGULAR LIFELONG VISITS WITH A PID SPECIALIST
(vary from every 3 months to every 2 years or more)

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

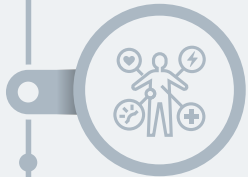
Processes and organisation for transitioning from paediatric to adult ward

Management of co-morbidities

Immunologists for adults or internists knowing PIDs/CVIDs

Identifying other specialists knowing PIDs

Follow up & Ageing Needs



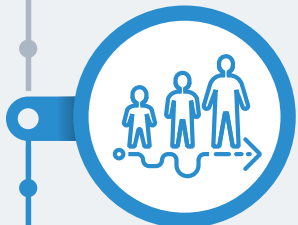
SYMPTOMS



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

Resources

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>

