POLICY RECOMMENDATIONS

WHEN RARE MEETS THE NOT-SO-RARE

STRENGTHENING EU ACTION ON PRIMARY AND SECONDARY IMMUNODEFICIENCIES

RECOMMENDATIONS AT A GLANCE

1. Strengthening early & accurate diagnosis

- Develop clinical guidance to help specialists identify underlying PIDs.
- Integrate screening into cancer pathways.
- Strengthen referrals across oncology, haematology, immunology and ERNs.
- Encourage recognition of immunology as a standalone specialty.
- Strengthen healthcare professional education on PIDs.

2. Ensuring equitable & sustained access to treatments

- Strengthen EU action to secure a sustainable supply of essential medicines and starting materials, including plasma, through the Critical Medicines Act.
- Ensure equitable access to immunoglobulin and essential treatments.
- Harmonise clinical guidance and eligibility criteria based on evidence and patient needs.
- Strengthen solidarity mechanisms to improve access to essential medicines.
- Coordinate EU action on antimicrobial resistance and vaccination for vulnerable populations.

3. Bridging rare disease & cancer policy frameworks

- Recognise immunodeficiencies as comorbidities in cancer survivorship and long-term care strategies.
- Establish integrated oncology–immunology pathways and multidisciplinary medical, nursing and psychological teams.
- Ensure continuity between paediatric and adult oncology and immunology services.
- Fund cross-disciplinary training through EU programmes.

4. Addressing psychological & social stigma

- Recognise specialised nurses and patient organisations as partners in long-term care and policymaking.
- Integrate immunodeficiency and psycho-social support into cancer survivorship care.
- Develop EU guidance addressing stigma, discrimination and invisible disabilities.
- Include mental health and quality-of-life indicators in EU health programmes and reporting.

5. Strengthening EU research, training & data collaboration

- Fund research into genetic predisposition and cancer-related immunodeficiencies alongside cross-disciplinary training.
- Support cross-border studies on disease progression and treatment outcomes.
- Enable secure cross-border health data sharing, respecting data protection requirements.
- Improve awareness of EU funding and simplify application processes.

INTRODUCTION

Primary immunodeficiencies (PIDs) are rare, mostly genetic disorders that impair the body's ability to fight infections. Secondary immunodeficiencies (SIDs), by contrast, are far more common and frequently arise as a consequence of cancer, autoimmune diseases, infections or their treatments.

While these immunodeficiencies differ in origin, their clinical manifestations frequently overlap. These findings highlight the need to rethink how immunodeficiencies are identified, managed and integrated into broader healthcare strategies. Especially because misdiagnosing a PID as a simple side effect of cancer treatment leads to higher long-term costs for health systems due to preventable hospitalisations.

The policy recommendations below were developed to ensure European health systems are equipped to address this inter-section effectively through the coordination of expertise, support for EU research and strengthening of healthcare resilience. It aims to ensure EU initiatives, such as the European Reference Networks (ERNs), the European Health Data Space (EHDS), the Critical Medicines Act (CMA), and the upcoming EU Rare Disease Action Plan, address gaps in immunodeficiency diagnosis, treatment access, and policy coordination across Europe.

Did you know...

Recent studies have shown that, in some patient cohorts, nearly 70% of patients initially diagnosed with SIDs may carry genetic variants associated with PIDs.[1]

STRENGTHENING EARLY & ACCURATE DIAGNOSIS ACROSS EUROPE

Many immunodeficiencies remain undiagnosed or are diagnosed late, particularly in EU Member States with limited specialised expertise and diagnostic capacity. In addition, in many EU countries, paediatric and adult clinical immunology subspecialty training programmes are not recognised or funded as standalone medical specialties.

As a result, patients with primary and secondary immunodeficiencies (PID and SID) are often managed by other specialists, such as pulmonologists, infectious disease specialists, haematologists, or rheumatologists, which can contribute to delays in recognising and diagnosing these conditions. Consequently, patients can wait many years before receiving an accurate diagnosis, delaying treatment and increasing the risk of severe complications.

Recent genetic research has revealed an important gap in current diagnostic approaches. A proportion of patients initially classified as having SIDs carry genetic variants associated with PIDs.[2] In many cases, patients who develop immunodeficiency following cancer treatment are never referred for genetic screening meaning underlying primary conditions may remain undetected.[3]

Policy recommendations

- **Develop EU-wide clinical guidance for oncologists, haematologists, and other relevant specialists to improve the identification of underlying PIDs.**
- **Promote immunodeficiency screening within European cancer care pathways, where clinically appropriate.**
- **Strengthen referral pathways between oncology and haematology services and the European Reference Networks (ERNs) to improve access to specialised, multidisciplinary care.**
- **Encourage Member States to formally recognise immunology as a standalone medical specialty, with adequate training, expertise, and access to specialised care.**
- **Strengthen healthcare professional education on recognising the warning signs of primary immunodeficiency to enable earlier diagnosis and**

> “It took me over 15 years to get diagnosed with a primary immunodeficiency. It has cost me so much. I’m scared for my future because no one thought of a primary immunodeficiency” [4]

ENSURING EQUITABLE & SUSTAINED ACCESS TO TREATMENTS

Patients living with immunodeficiencies often depend on specialised and/or prophylactic therapies, including immunoglobulin replacement therapy, targeted innovative therapies and biological therapies as well as on anti-infectious treatments. However, availability and access to these treatments varies significantly across Europe.

Supply shortages of plasma-derived medicines, including immunoglobulins, are well documented.[5] Some biologicals and innovative targeted therapies for the treatment of PIDs are not widely available in all EU countries. In addition, significant variation exists across and within countries regarding eligibility criteria and clinical guidance for access to immunoglobulin replacement therapy. As a result, some patients experiencing recurrent or severe infections may face barriers to treatment despite having a clear clinical need. Smaller EU Member States often face disadvantages in procurement negotiations, which can further restrict access to essential medicines.

Access to immunoglobulin replacement therapy is also increasingly relevant in the context of newer cancer treatments associated with a heightened risk of infection. In some cases, access to immunoglobulins may be an important component of the safe and effective use of these therapies. At the same time, rising antimicrobial resistance and vaccine hesitancy complicate infection management for vulnerable patients.

Policy recommendations

- **Strengthen EU action to secure a sustainable supply of essential medicines and starting materials, including plasma, through the Critical Medicines Act.**
- **Promote equitable access to immunoglobulin replacement therapy and other essential treatments across all EU Member States.**
- **Align clinical guidance and eligibility criteria for immunoglobulin therapy based on clinical evidence, patient needs, and infection burden.**
- **Strengthen cooperation and solidarity to improve access to essential medicines in smaller and resource-constrained Member States.**
- **Promote coordinated Europe-an action to tackle antimicrobial resistance and improve vaccination coverage and confidence among vulnerable populations, including people with immunodeficiencies.**

> “Infections and immunodeficiency are a daily challenge for me. I was diagnosed with myeloma in 2009 and have since undergone five lines of treatment, most recently CAR T-cell therapy. Since receiving CAR-T, the number of serious infections I have experienced has increased. All required hospitalisation and have had a major impact on my quality of life. Despite repeatedly requesting access to immunoglobulin therapies, I was informed that it was not available to me under current prioritisation criteria. Access to immunoglobulin therapies remains inconsistent despite clear clinical vulnerability.”
>
> *Myeloma patient, France*

BRIDGING RARE DISEASE & CANCER POLICY FRAMEWORKS

Despite their clinical overlap, rare disease strategies and cancer policies often operate in separate policy frameworks. Many healthcare systems remain organised around diseases rather than around the patient journey.

As a result, immunodeficiencies may be treated merely as side effects of cancer treatment rather than as conditions requiring specialised long-term management. Better coordination between rare disease frameworks and initiatives such as Europe's Beating Cancer Plan – which makes no reference to the relationship between cancer and immune conditions – is essential.

Policy recommendations

- **Recognise immunodeficiencies as relevant comorbidities in cancer survivorship strategies.**
- **Promote integrated oncology–immunology care pathways for coordinated patient management.**
- **Ensure structured transition pathways from paediatric to adult care to maintain continuity of oncology and immunology management.**
- **Support multidisciplinary teams integrating oncology, haematology, immunology, gastroenterology, dermatology, specialised nursing, psychological care, and other relevant specialties.**
- **Use EU programmes, including Horizon Europe and the European Social Fund, to support specialised training and strengthen cross-disciplinary expertise.**

ADDRESSING PSYCHOLOGICAL & SOCIAL STIGMA

For many patients, the transition from cancer survivorship to living with a chronic immunodeficiency can be psychologically challenging. Patients may expect to move beyond their illness only to discover that they require lifelong medical management. As survival improves for patients with conditions such as myeloma and chronic lymphocytic leukaemia (CLL), greater attention must be paid not only to survival, but also to living well while on treatment.

The impact of immunodeficiency extends far beyond physical health. Fear of recurrent infections can significantly restrict patients' daily lives, social participation and emotional well-being. Many patients report avoiding activities they previously enjoyed due to concerns about infection risk. Relationships with family members may also be affected as patients feel compelled to limit close contact to protect their health. These challenges can lead to social isolation, anxiety and a reduced quality of life.

This personal struggle is mirrored by a systemic failure in care. Across Europe, mental health services remain fragmented, with 65% of rare disease patients identifying the lack of psychological support as one of their primary challenges.[6] Healthcare systems are currently optimised for acute recovery rather than the long-term emotional resilience required for immunological conditions.

To bridge this gap, specialised nurses serve as a vital link, providing the expert treatment advice, education and emotional support patients need to navigate their "new normal." However, clinical care alone is not enough, the EU must address the societal barriers that follow these patients.

Primary and secondary immunodeficiencies are invisible disabilities. Because their challenges are often not visible to others, greater awareness and empathy are essential to creating understanding, providing appropriate support, and helping those affected manage their daily lives.

Policy recommendations

- **Recognise specialised nurses and patient organisations as essential partners in long-term immunodeficiency care, and strengthen their support at EU and national level.**
- **Ensure the meaningful involvement of patient organisations, clinicians, and nursing representatives in EU health policy development.**
- **Integrate immunodeficiency into cancer survivorship strategies, with appropriate long-term care and support for survivors.**
- **Develop EU guidance under the Expert Group on Public Health[7] to tackle discrimination and stigma, and advance the recognition of invisible disabilities.**
- **Promote the systematic inclusion of mental health and quality-of-life indicators in EU health programmes and the State of Health in the EU reports.**

> "After surviving two lymphomas, I believed the hardest part of my journey was behind me. Learning that I also had a primary immunodeficiency meant accepting a new reality – shifting from the hope of being cured to understanding that my immune system would always need protection."
>
> *PID patient*

STRENGTHENING EU RESEARCH, TRAINING AND DATA COLLABORATION

Research funding and healthcare investment remain fragmented across the EU, limiting the development of comprehensive research programmes and cross-border collaboration that are critical to addressing complex conditions such as immunodeficiencies.

Although healthcare delivery remains primarily the responsibility of EU Member States, the European Parliament's rare disease consultation revealed a concerning knowledge gap when it comes to EU funding opportunities for healthcare professionals and patient organisations.[8]

Policy recommendations

- **Support targeted EU research on the links between genetic predisposition and cancer-related immunodeficiencies, alongside cross-disciplinary training for immunologists, haematologists, and oncologists.**
- **Support longitudinal cohort studies across EU Member States to improve understanding of disease progression and treatment outcomes.**
- **Strengthen EU research collaboration by enabling secure cross-border data sharing in line with data protection requirements.**
- **Improve awareness of EU funding opportunities and streamline application processes to increase access to research funding.**

CONCLUSION

The growing scientific understanding of immuno-deficiencies highlights the need for a more integrated approach to patient care and health policy. The intersection between PIDs and SIDs illustrates how rare disease expertise can inform broader healthcare strategies.

By strengthening diagnostic capacity, improving treatment access, fostering multidisciplinary collaboration, and supporting research, the European Union can play a critical role in improving outcomes for patients across Europe.

Ensuring that these patients are not overlooked in broader health policies will require sustained political commitment, coordinated action across EU Member States, and the continued involvement of patient communities.

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