
Prevalence in the EU

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PID

PRIMARY IMMUNODEFICIENCIES

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Types of Primary Immunodeficiency

- B cell deficiencies
- T cell deficiencies
- Granulocyte deficiencies
- Complement deficiencies



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B Cell Deficiencies

- Hyper IgM syndrome (HIGM)
- Common variable immunodeficiency (CVID)
- IgA deficiency (IgAD)
- IgG subclass deficiency (IgGsD)



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Are PID Really Rare Diseases ?

- Rheumatoid arthritis - 1:100-200
- Gluten enteropathy - 1:200-800
- **Primary immunodeficiency - 1:250-500**
- Type 1 diabetes - 1:600-800
- Multiple sclerosis - 1:800-1000



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Diagnosed PID Patients in Sweden

• HIGM	11	(15-20)
• CVID	250	(400-500)
• IgAD	1 600	(15 000)
• IgGsD	500	(15 000 ?)
• C2D	41	(200-600)



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Expected PID Patients in the EU

- HIGM 600
 - CVID 25 000
 - IgAD 750 000
 - IgGsD 750 000 (?)
 - C2D 20 000
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- Sweden = 9 million, the EU = 460 million



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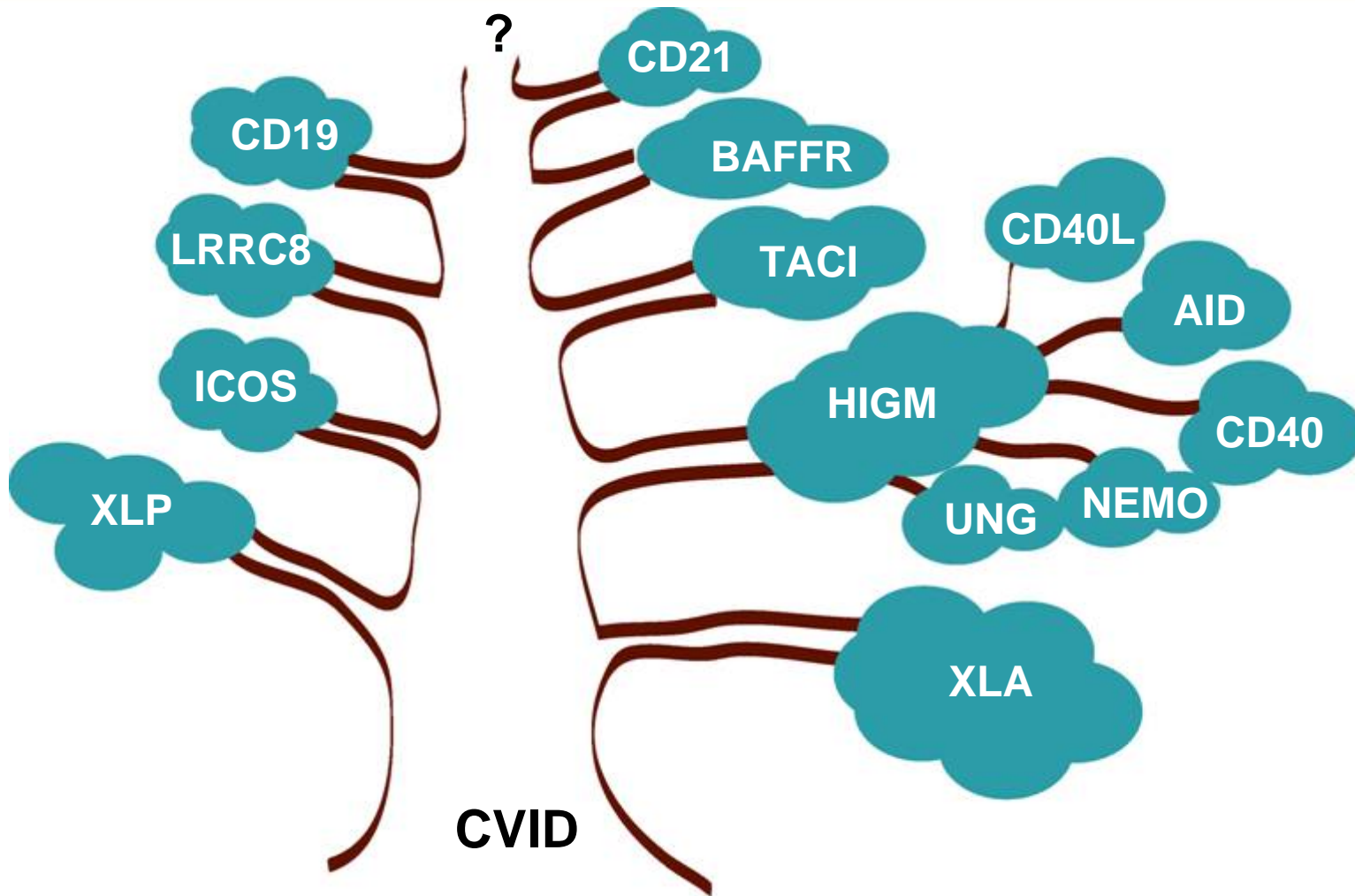
Diagnostic Problems/Potentials

- Awareness
- Diagnostic tools/centres
- Screening



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CVID



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Screening for PID

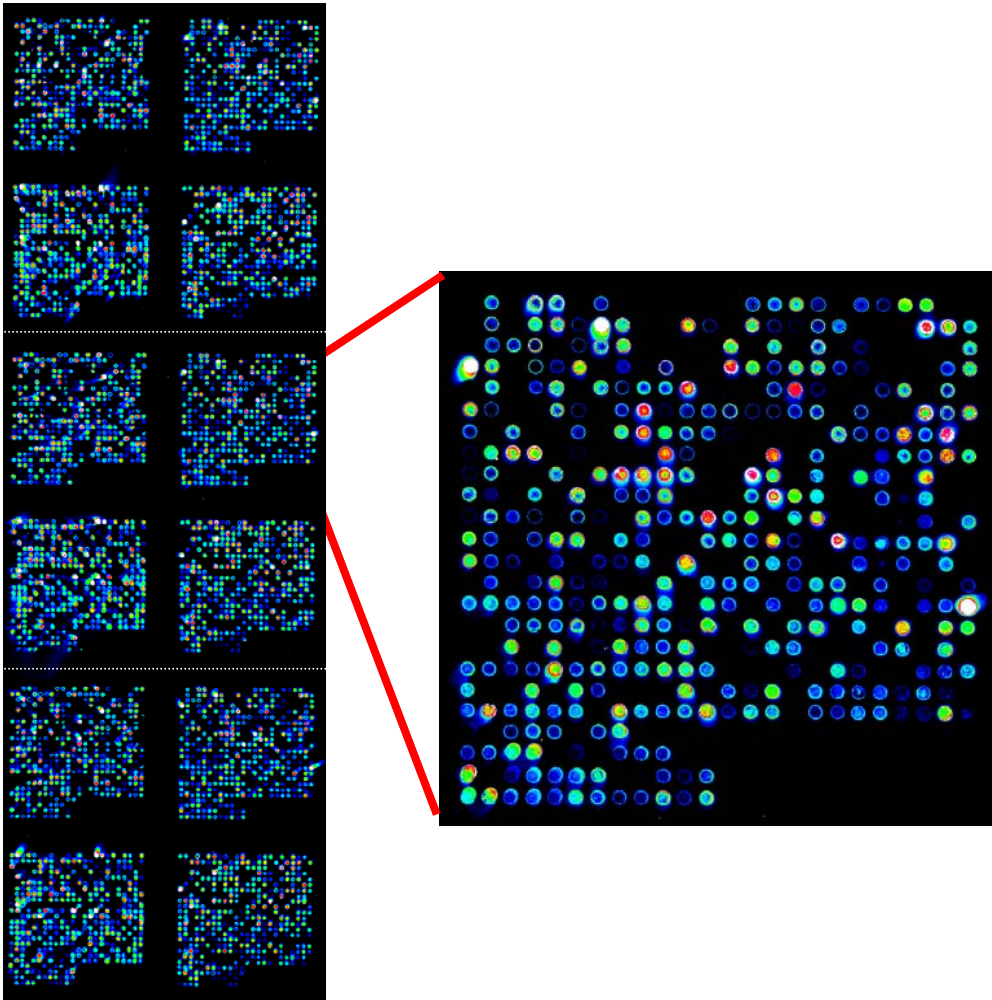
- **Population based**
- **Neonatal**



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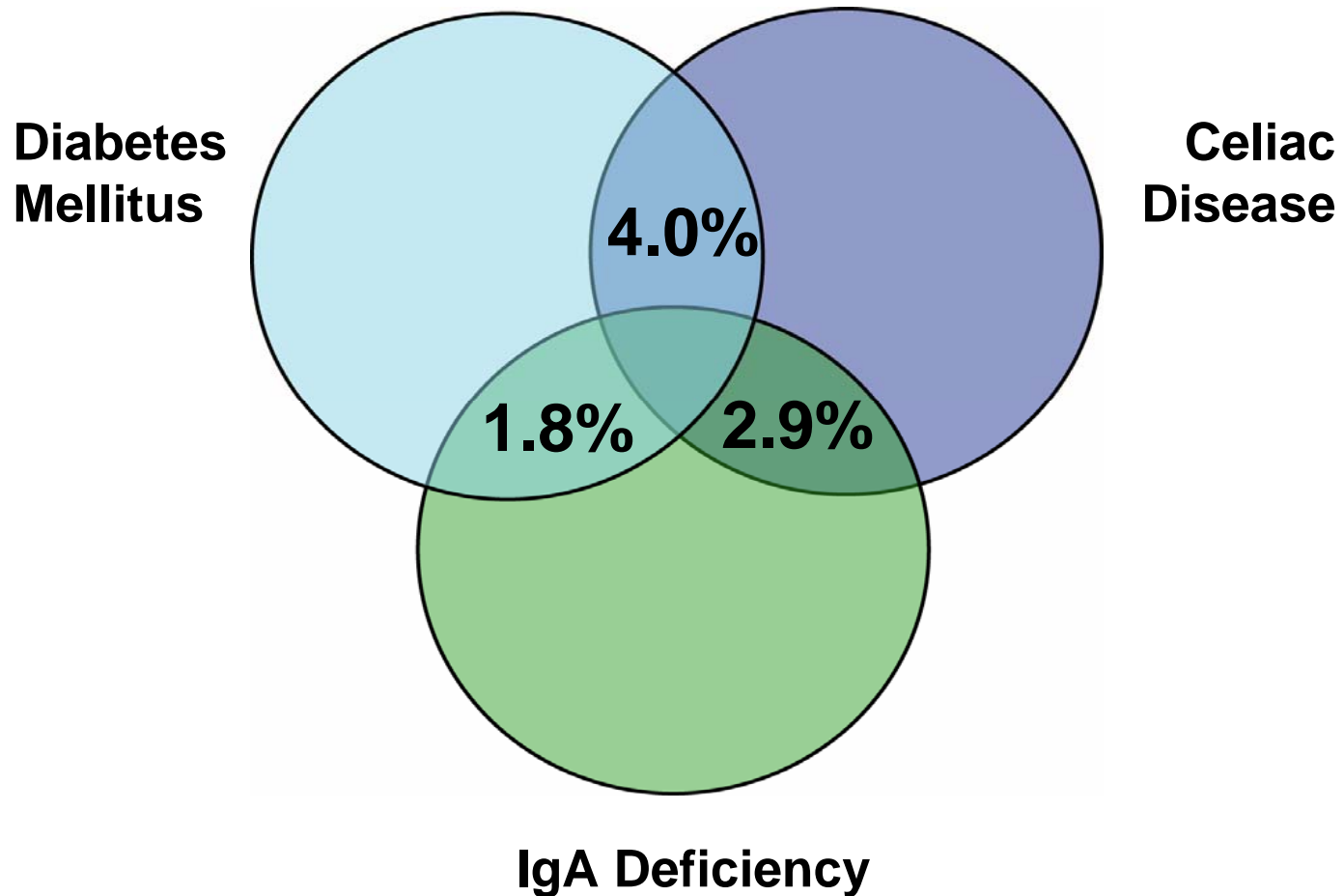
Reverse Array Screening for IgA



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PIDs Among Patients with Autoimmunity



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Screening for PIDs

- Population based
- Neonatal



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Screening Targets (PKU Samples)

- IgA (HIGM, CVID, IgAD etc)
- Complement (C2, C3)
- SNP based mutation analysis (SCID)
- T cell receptor excision circles (SCID)



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Patient Registries

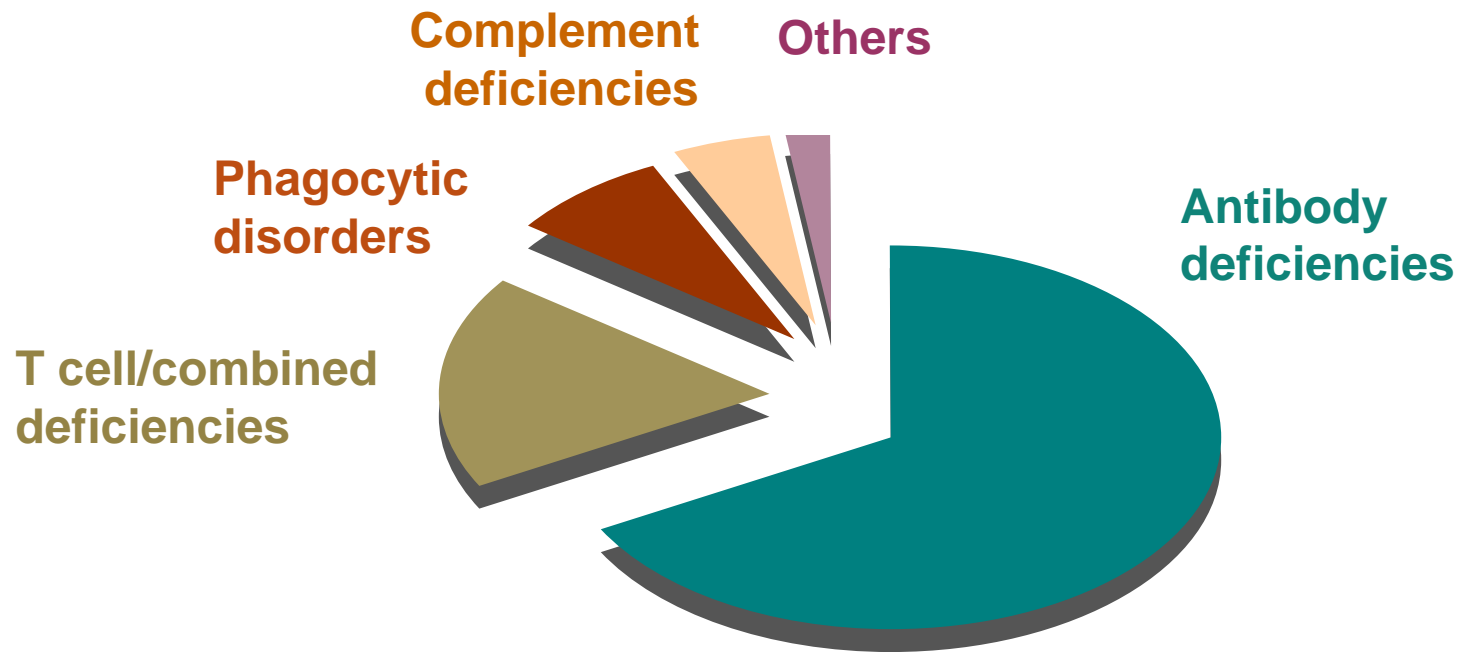
- **History**
- **Future**



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The “Historic” ESID Registry



Total number of patients recorded in the registry: 10 231



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Patient Registries

- History
- **Future**



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Online ESID Registry

