

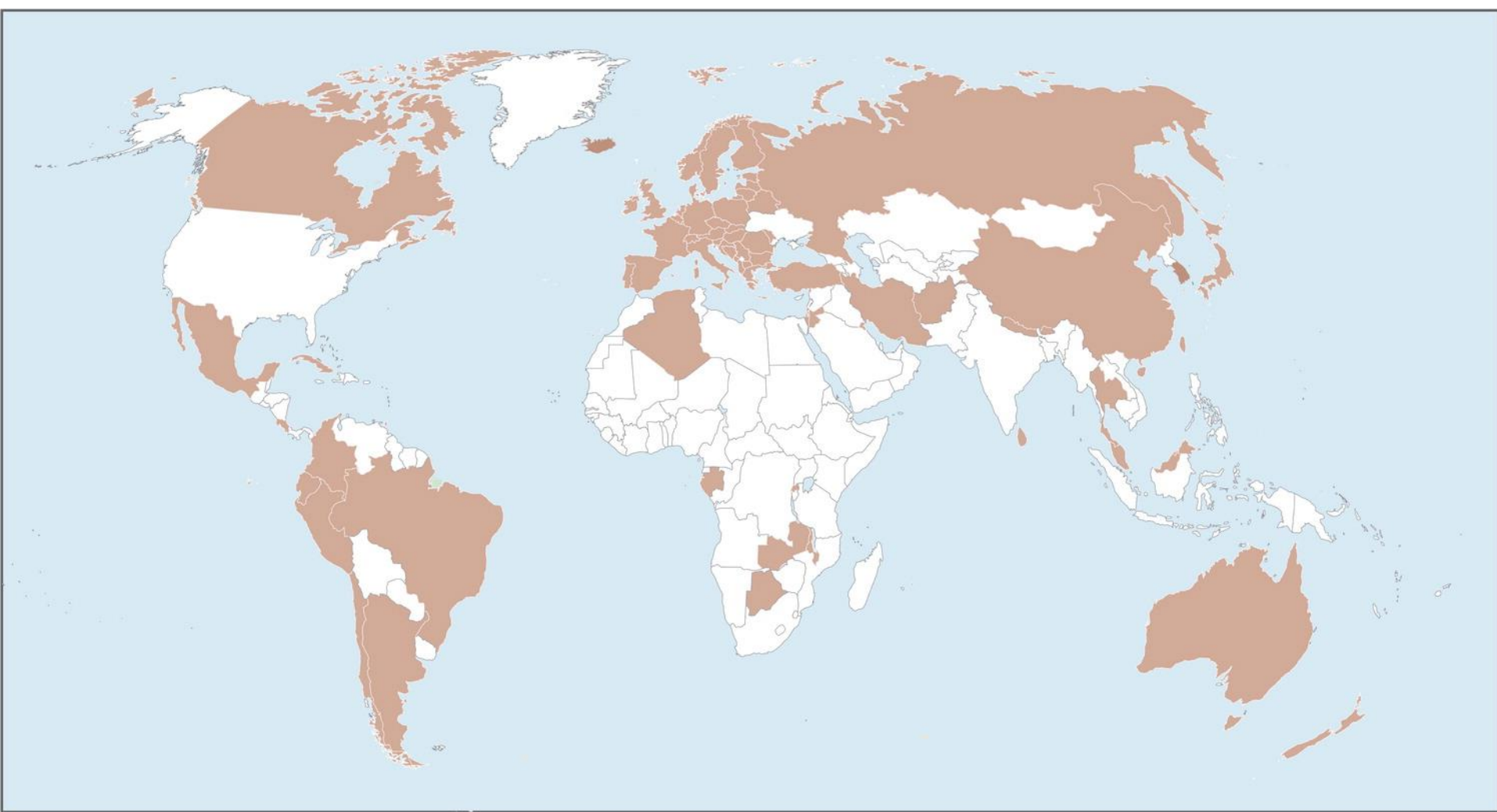
Role of specialized PID centres & the importance of registries

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Countries That Have Adopted Reforms toward Universal Health Coverage.



Primary immunodeficiency disease

- Specialized PID centres
- PID Registraties

If not treated, can be chronic, life-long, serious, and even fatal

Criteria for fast and reliable PID diagnoses

- **Early recognition of clinical manifestations**
- **High-profile medical awareness of PIDs and information campaigns for referral of the patient**
- **Consensus on basic screening tests, available to all primary health care and hospital doctors (i.e., complete blood count and differential; quantification of serum Ig levels)**
- **Immediate access to a PID specialist for confirmation of diagnosis and speedy treatment**
- **Standardization of immunological diagnostic protocols**
- **Access to genetic counseling for the patient's family after diagnosis**



Specialized Immunodeficiency Centre

Specialized diagnostic laboratory



- Excellent clinician/nurses team
- Experienced laboratory support or closely connected
- Interaction with patient organisations
- Participation in regional/national/international network
- Education/training centre
- Research (participation)

Pediatrics

Infection/Immunology

Transition



Internal Medicine

Clinical Immunology

Managing the “Burden of Disease” in PID

- Infections
- Persistent organ damage, for example lungs
- Autoimmunity/malignancy
- Burden of treatment for infections/autoimmunity/other complications

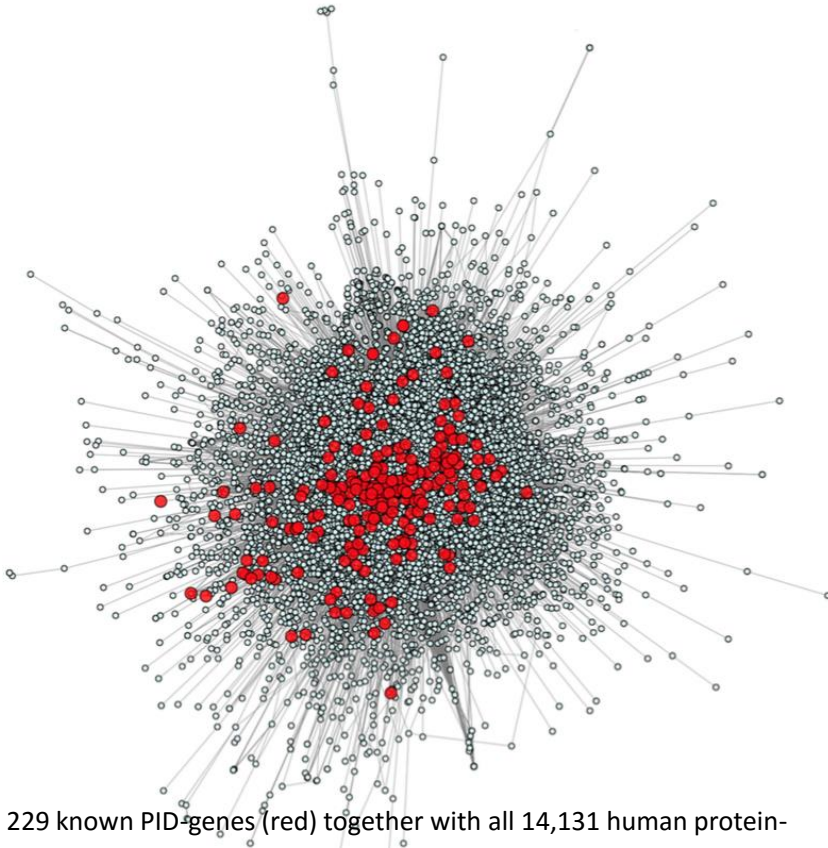
Key disciplines related to PID services

- **High-quality laboratory diagnostic services, including histopathology**
- **Radiology services**
- **Intensive care services**
- **Infectious diseases consultancy**
- **Respiratory medicine**
- **Clinical hematology, including transplantation**
- **Gastroenterology/hepatology (adult and pediatric), including endoscopy**
- **Dermatology,**
- **Other tertiary services may also be needed, for example, otorhinolaryngology (ENT), as well as social services and psychiatry**
- **Clinical genetics**

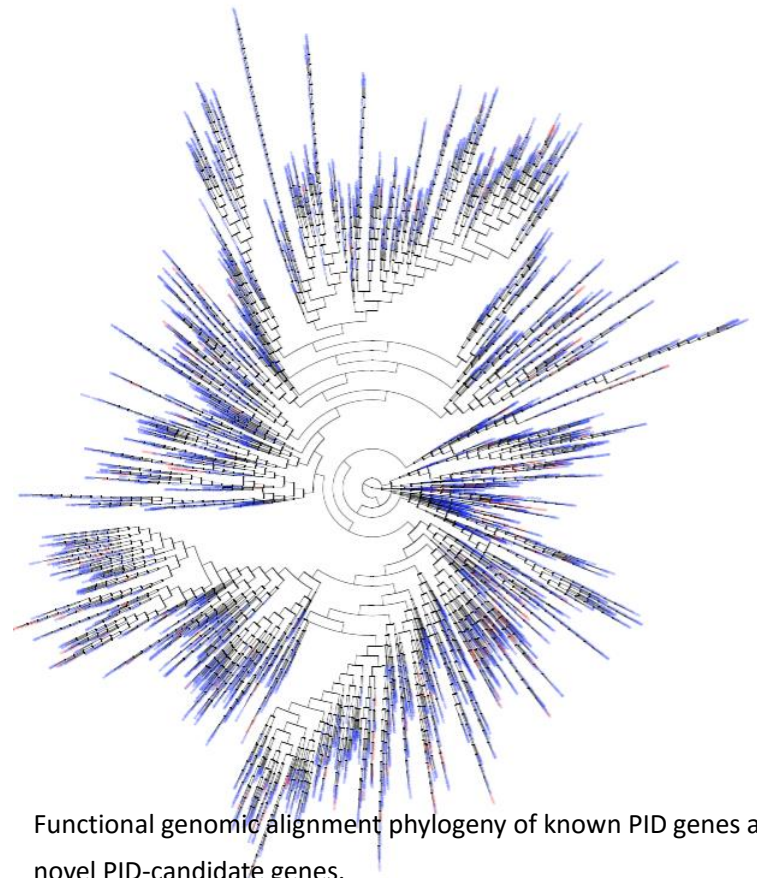


Because!!

At the moment > 260 genes are known in PID



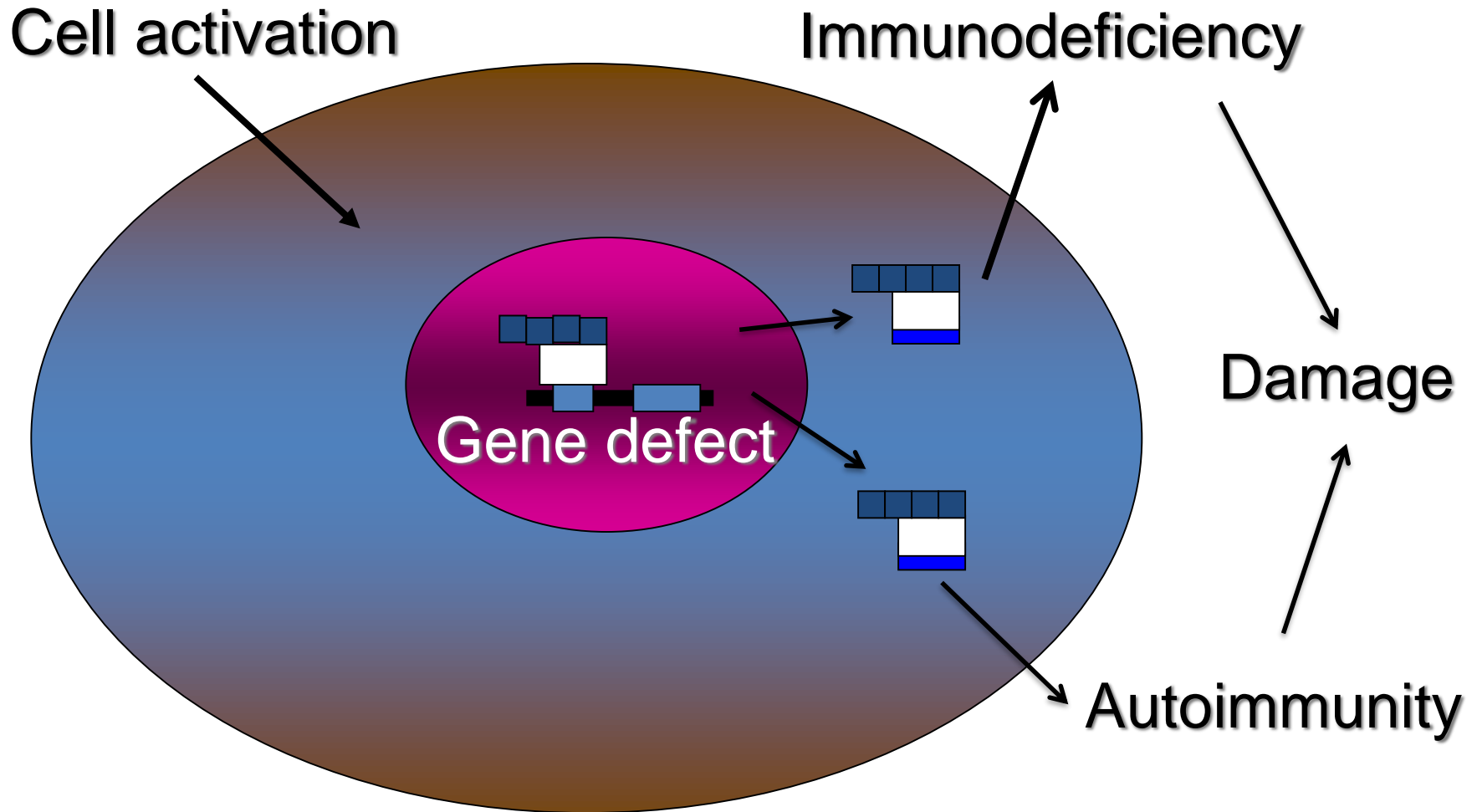
229 known PID-genes (red) together with all 14,131 human protein-coding genes for which HGC-predicted biological distance



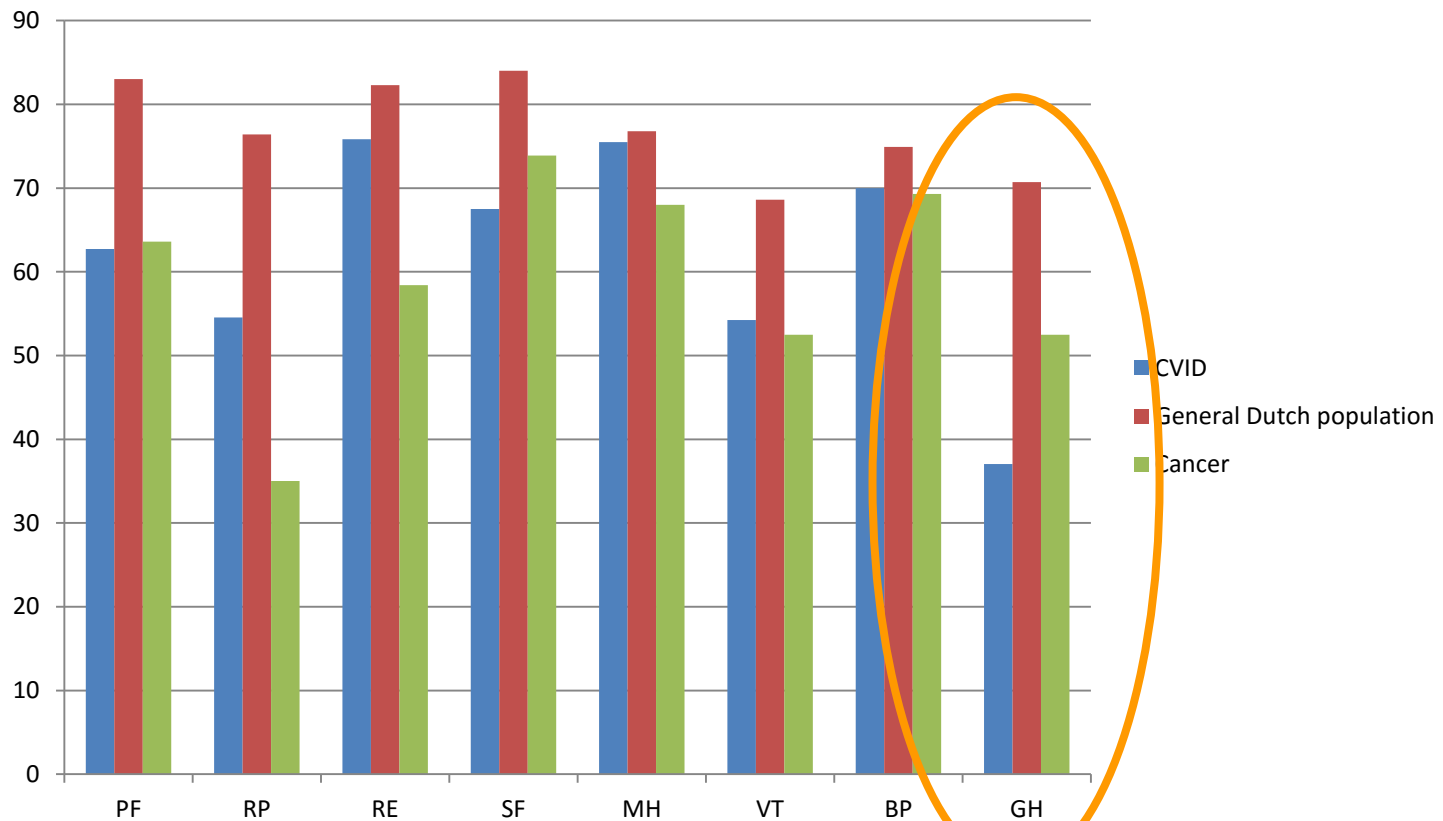
Functional genomic alignment phylogeny of known PID genes and novel PID-candidate genes.

Gene defects in PID

one defect 2 diseases



Quality of Life: Means for SF-36 dimensions of CVID, General Dutch population and Cancer



PF=Physical Function, RP=Role Physical, RE=Role Emotional, SF=Social Functioning, MH=Mental Health, VT=Vitality, BP=Bodily Pain, GH=General Health.

Van der Ent, not published

Why PID registrations?

Levels of information

- level 1 encompasses data fields that are mandatory for all who are registered, thus enabling **epidemiological studies**
- Levels 2 and 3 are optional, with level 2 including **diagnostic and follow-up data**
- level 3 will comprise projects running for limited time-periods to collect data for use in **clinical studies**

National registries

- National registries are important tools for;
 - Incidence
 - Prevalence
 - Detection of areas of low-diagnostic rate
 - Provision of insights on diagnostic delay
 - Morbidity and mortality

National registries

- Helpful to governments/health policymakers/health insurers regarding estimates;
 - of those not diagnosed
 - planning of educational programs
 - provision of treatments and their costs
 - **Presentation of this data to pharmaceutical industries helps to ensure that the supply of relevant medical products meets demand**

What Can We for Example Offer Our PID Patients?^{1,2}

Unfortunately “Oral IgG” is out of the question!

But we have efficacious and well-tolerated therapy options:



IVIG



SCIG

Proven efficacy in PAD

Issues;
Quality
Availability
Reimbursement
Exchangeability

1. Espanol T, et al. *Patient Pref Adherence*. 2014.

2. IPOPI Patient Needs & Outlooks Survey. Available from: http://www.ipopi.org/uploads/IPOPI%20PID%20Patient%20Survey%20-%20Shortened%20Report%20-%20030812_Final%20IPOPI%20format.pdf. Accessed 25 August 2014.

International networks

- **IPOPI!**
- **Latin American Society for Immune Deficiencies (LASID)**
- **African Society for Immune Deficiencies (ASID)**
- **South-East Asian Primary Immunodeficiency network (SEAPID)**
- **European Society for Immune Deficiencies (ESID)**
- **United States Immune Deficiency Network (USIDNET)**

International registry networks

- **Collect information on all PIDs including the very uncommon cases**
 - **ESID set up an initial registry in 1994 in Sweden, supplemented by an online database in 2004**
- **Last 2 years, the ESID registry has been completely revised in order to provide information for national registries, healthcare providers and the European Commission**

In conclusion

- National PID expert centres are the mainstay in the regional diagnosis and treatment of PIDs
- Rare diseases need registration especially in case of multiple symptoms, comorbidities and limited treatment possibilities to optimize patient treatment
- Learn to prevent organ damage!!
- Personalized medicine in PID may be thought of as the tailoring of medical treatment to the individual characteristics, needs, and preferences of a patient during all stages of care (adapted from the FDA)¹