

# ITALIAN NETWORK FOR PRIMARY IMMUNODEFICIENCIES (IPINET): AN USEFUL OPERATIVE MODEL FOR RARE DISEASES



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## Summary

The IPINET web-based system is an interesting operative model applied for rare diseases research and clinical care. It is a national network of Centers of expertise and non-specialized centers, that produces, shares and updates disease-specific diagnostic and therapeutic protocols, improving the quality of care and the quality of life for patients with Primary Immunodeficiencies

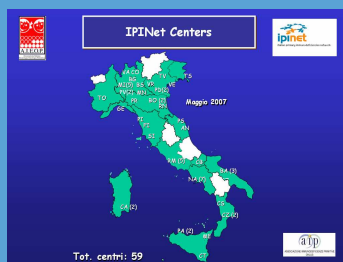
## Introduction and aims

The management of Primary Immunodeficiency Diseases (PIDs), rare diseases due to defects of the immune system, presents difficulties concerning assistance organization, clinical care and research. To overcome that, the IPINET, established in 1999 within the Italian Association of Pediatric Hematology and Oncology and with the support of the Italian patients Association of Primary Immunodeficiencies (AIP), aims to:

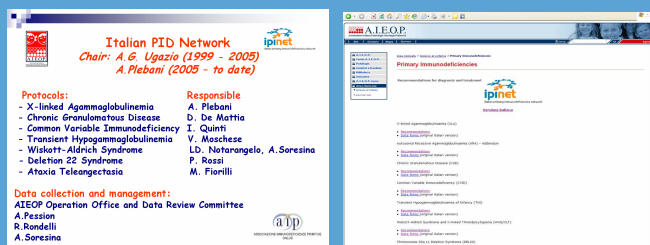
- assure a definitive molecular diagnosis to all cases with clinically diagnosed or suspected PIDs by means of highly qualified referral laboratories;
- decrease health migration and its related individual and social costs by means of a network of Centers of expertise and non-specialized centers, that produces, shares and updates disease-specific diagnostic and therapeutic protocols;
- build a centralized system for PIDs data collection to evaluate both patient accrual and the long-term efficacy and late-effects of previously and currently adopted treatments;
- share knowledge and experience between centers participating in the web network program.

## Methods

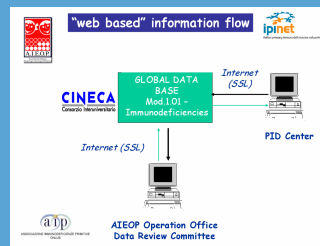
Fifty-nine Centers were identified including non-specialized hospitals and centers highly specialized in PID.



Representatives from these centers have jointly formulated and adopted common protocols for diagnosis and treatment of children and adults with X-linked and Autosomal recessive Agammaglobulinemia, Chronic Granulomatous Disease, Common Variable Immunodeficiency, Transient Hypogammaglobulinemia, Wiskott Aldrich syndrome, Deletion 22 syndrome and Ataxia Teleangectasia, available on Italian web site [www.aieop.org](http://www.aieop.org); an english version is available linked to the ESID web site, [www.esid.org/links](http://www.esid.org/links).



The IPINET identified referral laboratories for molecular diagnosis and utilized a web-based centralized system for data collection and analysis of the PIDs at the Italian Interuniversity Computing Center (CINECA). The system allows management of the whole informative flow with consultation of protocols and exchange of information across forum. Each center enters patients' information by electronic forms of registration, diagnosis, therapy, side effects, annual follow-up.



## Results and Conclusions

From 1999 to the present day, the IPINET has:

- optimized operator work,
- improved the ability of physicians to manage such a rare diseases,
- obtained the enrolment of large series of cases (123 XLA, 30 AAR, 68 CGD, 303 CVID, 85 THI, 59 WAS, 103 Del22), assuring a large amount of high quality data, and
- lead to an important amelioration of both clinical assistance and quality of life for PIDs children and adults.

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