



Piedmont and Valle d'Aosta

Interregional Network for Rare Diseases

Piedmont and Valle d'Aosta Registry of Rare Diseases

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**Coordinamento della Rete Interregionale per le Malattie Rare del Piemonte
e della Valle d'Aosta**



Background

The Piedmont and Valle d' Aosta Interregional Registry of Rare Diseases is part of the National Network for Rare Diseases of the Italian Institute of Health.

Aim of the project:

- Collecting data on the prevalence of a wide deal of rare diseases
- Developing diagnostic and therapeutic protocols
- Providing patients, associations, and health workers with information about rare diseases.
- Allowing a rational distribution of funds

Decentralized model

Critical role of the network
Reference centers only for diagnosis
Promotion of working groups
Elaboration of consensus statements
Attempts to assure equal assistance



Law about Rare Diseases in Italy, Piedmont and Valle d'Aosta

DM 279/2001: the key national law about rare diseases.

D.G.R. 22-11870: Piedmont regional law promulgated in 2004

- Regional Network for prevention, epidemiologic surveillance, diagnosis and therapy of rare diseases that includes each Public Health Facility of the region;
- Network coordinated by the “Centro Multidisciplinare di Ricerche di Immunopatologia e Documentazione su Malattie Rare (CMID)”, which holds the Regional Registry of Rare Diseases, promotes medical education and cooperates with the Italian Institute of Health and the official Italian institutions and patient associations.

D.G.R. 38-15326: Piedmont regional law promulgated in 2005

- Extends to other 40 pathological disorders the benefits (diagnostic tests and best treatment) attributed to the diseases listed in DM 279-2001
- Institutes a committee of technical and medical experts to support the coordinating Centre

D.G.R. 234-2008: Valle d'Aosta regional law promulgated in 2008

D.G.R. 21-8414-2008 Piedmont regional law promulgated in 2008

- Create an interregional network for rare diseases

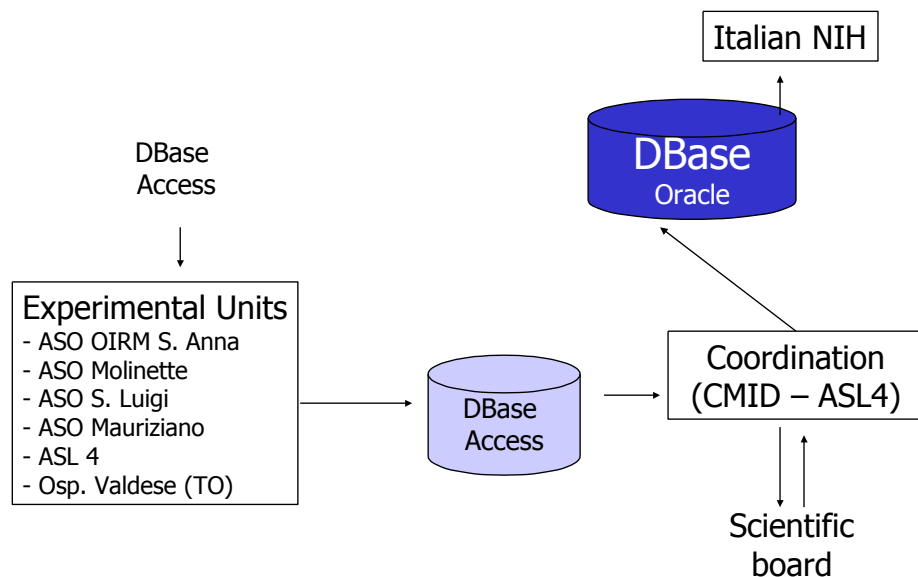


Development of the Registry

- First phase:
 - lasted 6 months
 - 6 Experimental Centers.
 - Data dealing with 832 patients
- Second phase:
 - started in January 2006 and currently ongoing
 - Attempt to involve every Public Health Facility in Piedmont and Valle d'Aosta in the collection of data
 - Database for data collection and processing developed by the Regional Agency of Informatics (CSI)

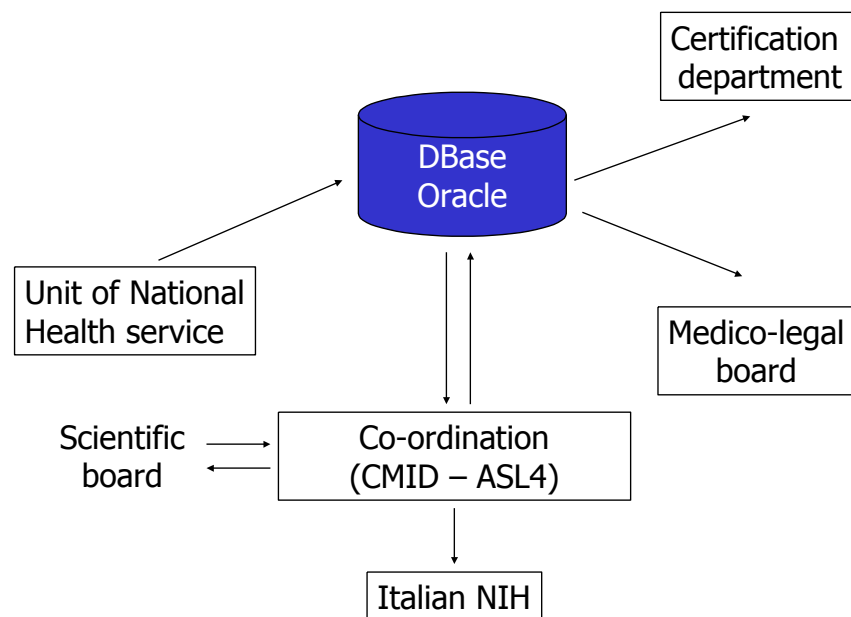


Experimental phase



During the experimental phase 832 cases were registered according to the data collection form suggested by the Italian National Institute of Health

Present organization





Data collected

PATIENT PERSONAL DATA

- Vital statistic data
- Social data

DISEASES RELATED DATA

- Onset
- Diagnosis
- Sign and symptoms

DISEASE DIAGNOSTIC CRITERIA

- Clinical criteria
- Laboratory criteria
- Instrumental criteria

THERAPEUTIC PRESCRIPTIONS

- Compelled link to registration form
- Extensive administration of innovative drugs, galenics for life-threatening conditions, off-label medications

The screenshot displays a medical software interface titled "Malattie rare - Registro regionale dei pazienti affetti da malattie rare". The interface is divided into several sections:

- Paziente:** A sidebar menu with options like "Archivio", "Scheda Malattia Paziente", "Scheda Criterio Diagnostico", "Piano terapeutico", "Archivio Malattie", "Pazienti", and "Gestione Utenti".
- Scheda malattia paziente - [Definitiva]:** The main form for patient data, including:
 - Dati identificativi:** Fields for Cognome, Nome, Data di nascita, Sesso, Cittadinanza, and Codice fiscale.
 - Lungo di nascita:** Fields for Stato, Provincia, Comune, and Comune - Località straniera.
- Scheda malattia paziente (modalità):** A small dialog box for selecting the type of patient record (Definitiva, Storica, Temporanea).
- Ricerca piani terapeutici:** A search window for therapeutic plans, including fields for Paziente (Cognome, Nome, Data di nascita, Codice fiscale) and Piano Terapeutico (ID PT, Stato PT).
- Malattia:** Fields for Codice, Sub. cod, and Nome malattia.
- Farmaco:** Fields for Tipo farmaco and Farmaco.
- Stato:** A section for "IN MODIFICA" with fields for Inserita da and Modificata da.

The bottom of the screen shows a Windows taskbar with the "start" button and several open applications, including "Malattie rare - Regist..." and "Adobe Reader - [con...]". The system clock indicates the date is 11/10/2015 and the time is 14:10.



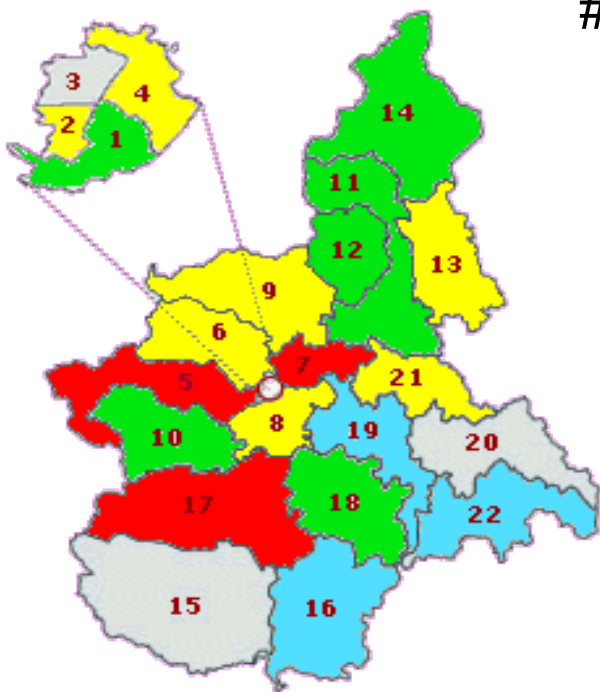
Comparison with other pathological conditions

- **Diabetes** is a disease that impairs life expectancy and is associated with chronic debilitation. It requires a peculiar joined effort to avoid a significative morbidity and a premature death...
- **CODE-2 study** evaluated direct and indirect health costs and social costs for pts affected by type 2 diabetes in Italy → 3.079 € / year for each pts
- **Turin** inhabitants with social benefits because of diabetes¹ → 31,700
- **Estimated yearly cost for diabetes in Turin:** 97,813,672 €
- **Piedmont** inhabitants with social benefits for rare diseases → 17,500
- **Estimated yearly cost for rare diseases in Piedmont:** 17,505.644€

1) Data at **February 2007**; Analysed population: 901.010



Estimated prevalence of rare diseases in different areas of Piedmont region



/100,000 inhabitants

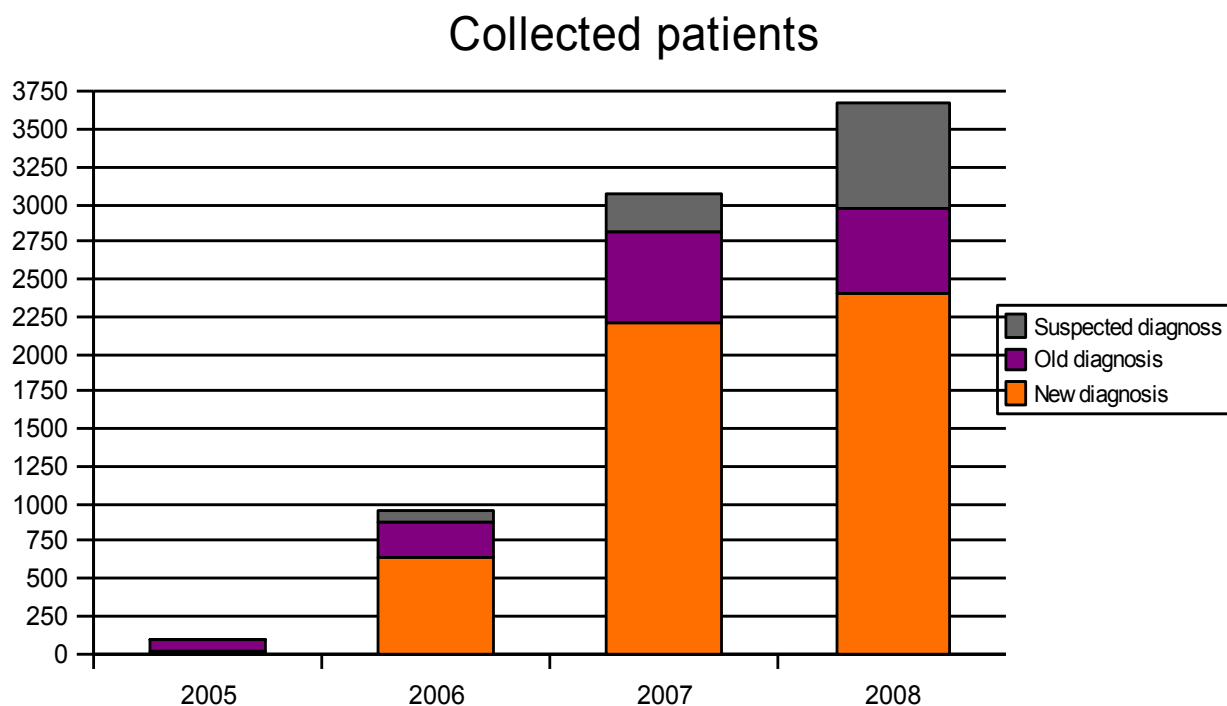
< 100	Light Blue
100 – 149	Green
150-200	Yellow
> 200	Red

Since DM 279/2001 until 2008 about 22,400 pts recognized as being affected by rare diseases and provided with a certification for free access to medical assistance in Piedmont



Dec 31th 2009

- A total of 6773 cases have been recorded in the registry
- 1037 cases waiting for confirmation





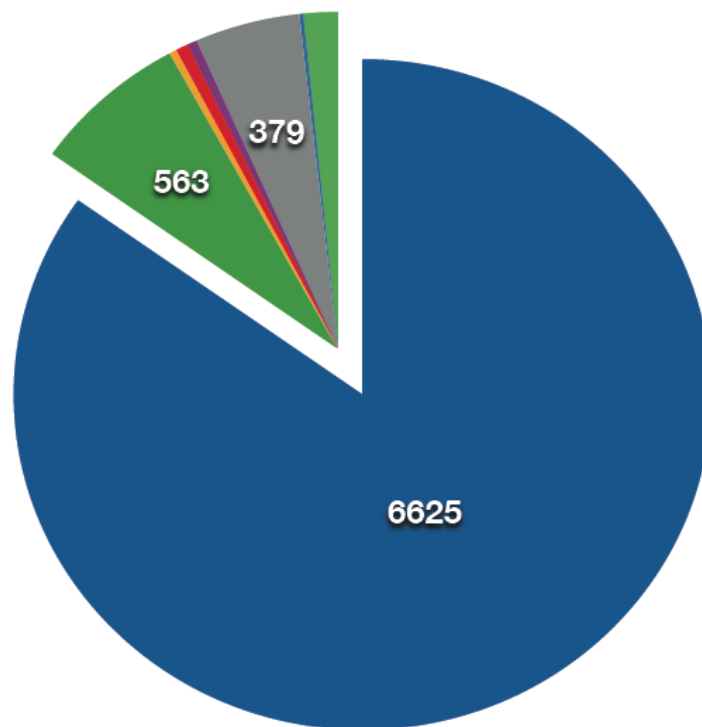
	Suspected diagnosis	Established diagnosis	Total
INFECTIOUS AND PARASITIC DISEASES (001-139)	49	145	194
NEOPLASMS (140-239)	42	337	379
ENDOCRINE, NUTRITIONAL AND METABOLIC DISEASES, AND IMMUNITY DISORDERS (240-279)	128	1020	1148
DISEASES OF THE BLOOD AND BLOOD- FORMING ORGANS (280-289)	233	1384	1617
DISEASES OF THE NERVOUS SYSTEM AND SENSE ORGANS (320-389)	158	1060	1218
DISEASES OF THE CIRCULATORY SYSTEM (390-459)	82	289	371
DISEASES OF THE RESPIRATORY SYSTEM (480-519)	38	139	177
DISEASES OF THE DIGESTIVE SYSTEM (520-579)	19	284	303
DISEASES OF THE GENITOURINARY SYSTEM (580-629)	36	97	133
DISEASES OF THE SKIN AND SUBCUTANEOUS TISSUE (680-709)	6	196	202
DISEASES OF THE MUSCULOSKELETAL SYSTEM AND CONNECTIVE TISSUE (710-739)	147	799	946
CONGENITAL ANOMALIES (740-759)	98	1020	1118
CERTAIN CONDITIONS ORIGINATING IN THE PERINATAL PERIOD (780-779)	0	2	2
SYMPTOMS, SIGNS, AND ILL-DEFINED CONDITIONS (780-799)	1	1	2
Total	1037	6773	7810



Diseases or groups	# of patients
Coagulation hereditary diseases	566
Hereditary anemia	495
* Scleroderma	472
Neurofibromatosis	322
ALS	320
Aminoacid metabolism diseases	204
Down syndrome	200
UCTD	193
Celiac disease	182
Muscular dystrophy	169
* APS syndrome	158



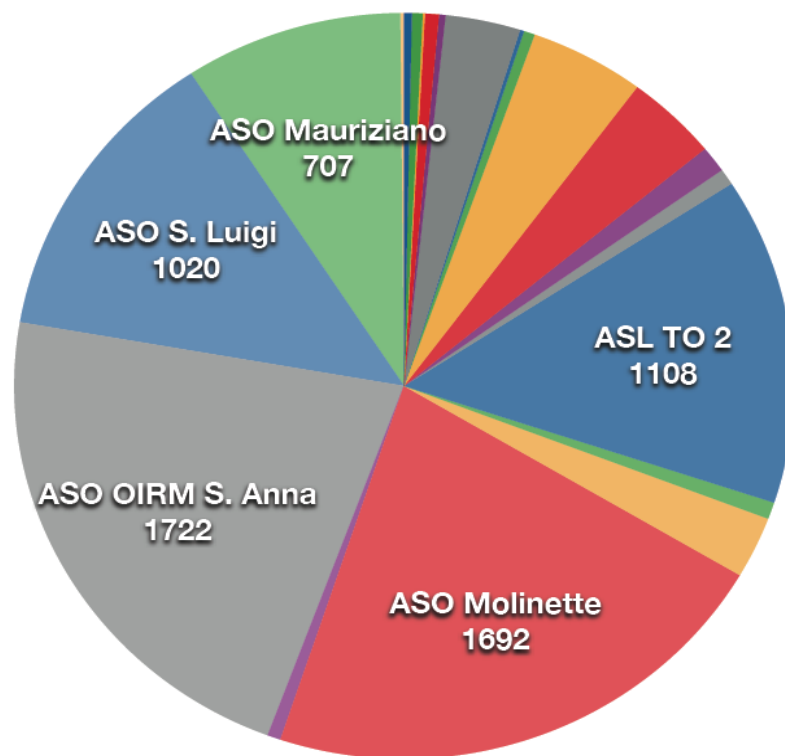
Patients file combined by health district that signaled the case



- Turin District
- Vercelli district
- Biella district
- Asti district
- Cuneo district
- VCO district
- Novara district
- Alessandria district



Patients Case divided by signaling health facility



- | | | |
|---------------|-----------------|------------------|
| ASL Vercelli | ASL Biella | ASL Novara |
| ASL VCO | ASL CN 1 | ASL CN2 |
| ASL Asti | ASL Alessandria | ASO Novara |
| ASO Cuneo | ASO Alessandria | ASL TO 1 |
| ASL TO 2 | ASL TO 3 | ASL TO 4 |
| ASO Molinette | ASO CTO | ASO OIRM S. Anna |
| ASO S. Luigi | ASO Maurizioano | Presidi |



Piedmont and Valle d'Aosta Consortia for Rare diseases

Rational use of resources

Evaluation of the efficacy of different therapeutic algorithms

Increase of direct drugs distribution by Public Health facilities

- Pulmonary hypertension
- APS
- Amyloidosis
- Prader Willi Syndrome
- Lysosomal diseases
- UCTD
- Systemic sclerosis
- Porphyria
- Bone dysplasia
- Huntington's disease and related conditions
- Arnold-Chiari and syringomyelia
- Autoimmune neuropathies
- Precocious puberty



RETE REGIONALE

Malattie Rare

[Home - ricerca](#) / [Rete assistenziale](#)



Sezioni

[Home page - ricerca](#)

[Legislazione sanitaria](#)

[Legislazione socio
assistenziale e previdenziale](#)

[News e convegni](#)

[Rete assistenziale](#)

[Associazioni di Volontariato](#)

[Link ad altri siti](#)

[Dichiarazione di accessibilità
e tasti di accesso](#)

Visualizzazione

[Imposta testo piccolo](#)

[Imposta testo medio](#)

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Rete assistenziale

Le **ASL** sono delle **Aziende Sanitarie** che forniscono servizi di tipo diffuso sul territorio e, a volte ma non sempre anche servizi di tipo ospedaliero. In Piemonte sono presenti **22 ASL** che sono ulteriormente suddivise in distretti. Nell'ambito delle malattie rare le ASL si fanno carico dell'assistenza locale dei pazienti e dell'erogazione dei farmaci, tramite le proprie Farmacie Ospedaliere; oltre alle aziende sanitarie locali sono coinvolte nella rete regionale piemontese malattie rare le **Aziende Sanitarie Ospedaliere** e le **I.R.C.C.S.**

[ASO Maggiore della Carità](#)

[ASO O.I.R.M.-St.Anna](#)

[ASO Ordine Mauriziano](#)

[ASO S.Giovanni Battista](#)

[Azienda Ospedaliera Alessandria](#)

[Azienda Ospedaliera C.T.O.- C.R.F. ICORMA](#)

[Azienda Ospedaliera S. Croce e Carle](#)



Website access

- 56577 visitors in 2007 and 70776 in 2008 accessed the site
- Mean daily number in 2008: 193 visitors

	2008	2007
January	6875	1275
February	6124	5416
March	5523	5841
April	5754	5532
May	6461	5904
June	6553	5200
July	5801	4907
August	4127	3978
September	6170	4561
October	6643	5150
November	6090	4630
December	4655	4183
Total access in the year	70776	56577
Mean daily access	193,38	155,01



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