
Feed Back of RSG Meeting Japan 2007

Robert Jakob, WHO



Steps

- Press Conference
 - Video Message ADG Dr T Evans
 - Note Verbale DG Dr M Chan
 - Press Release
- Meeting with Japanese Scientific Societies
 - 29 Representants
 - Chair Dr Fujiwara
 - MoH

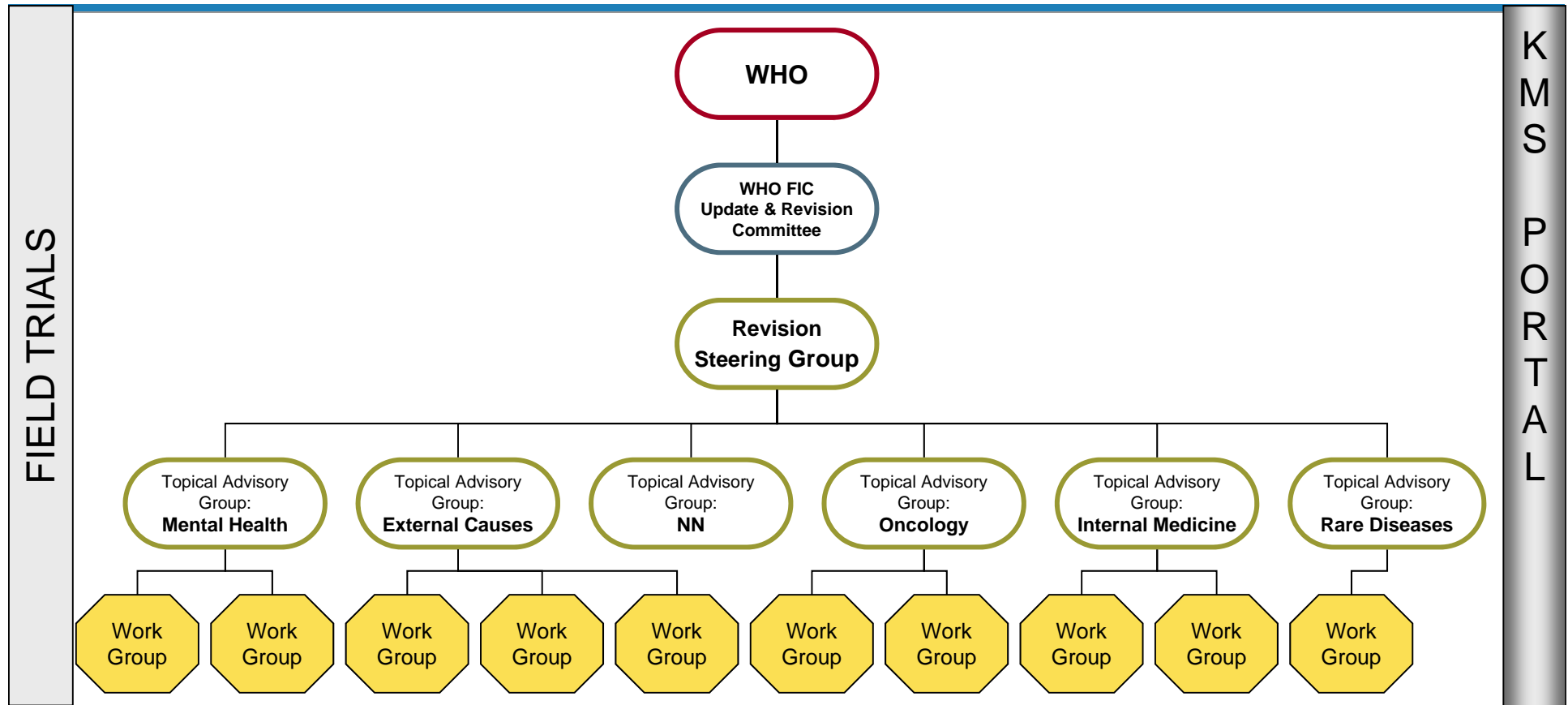


Steps

- Day 1:
 - Revision Process
 - Technical Platform
 - Presentations of the Topical Advisory Groups
- Day 2:
 - Taxonomy, ontology, linguistic issues
 - Partnerships
 - Coordination

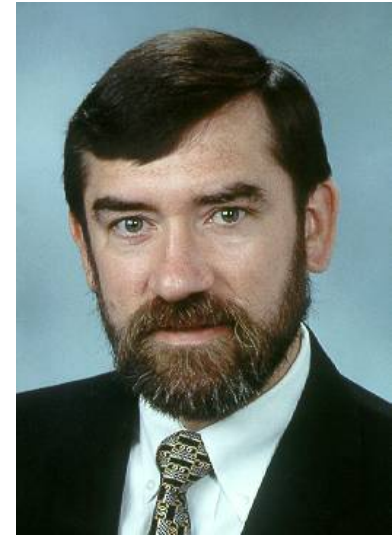


ICD-10 Revision organization structure



The Revision Steering Group

Chair of the RSG



Christopher G Chute

Mayo Clinic
Rochester, USA



The Revision Steering Group

Rare Diseases

Chair: Ségolène Aymé

Orphanet

Rare Diseases Platform

Broussais Hospital, Paris, France



The Revision Steering Group

Injury and External Causes of Injury

James Harrison

AIHW National Injury Surveillance Unit
Flinders University – Adelaide – South Australia

& WHO Dpts of :

- Violence and Injury Prevention
- Drug safety
- Occupational Health



The Revision Steering Group

Mental Health

Chair: Steven E Hyman

Provost

Harvard University, Boston, USA

& WHO Dpt Mental Health



The Revision Steering Group

Internal Medicine



Chair: Kentaro Sugano

Department of Internal Medicine
Jichi Medical University, Japan

& WHO Dpt Non Communicable Diseases



The Revision Steering Group

- **Marjorie S. Greenberg**
- **Chair of the
Planning Committee
WHO-FIC**



- **Martti Virtanen**
- **Chair of the
Terminology Reference Group
WHO-FIC**



The Revision Steering Group

- **Richard Madden**
- **Chair of the Family Development Committee WHO-FIC**

- **Mea M. C. Renahan**
- **Chair of the Update and Revision Committee of the WHO-FIC**



Cancer

- collaboration with IARC:
 - Blue Books (“Classification of Tumours”)
 - Explore compatibility (ontology, genes etc)
- collaboration with WHO team Cancer Surveillance
 - Needs: e.g. HPV - Cervical Cancer
 - Partners: UICC, FIGO ...
- ICD
 - Alignment with ICD-O
 - Sample of ICD-11 for Ch. II, breast cancer
 - Reviewing structure
- Setting up a working group in collaboration with IARC

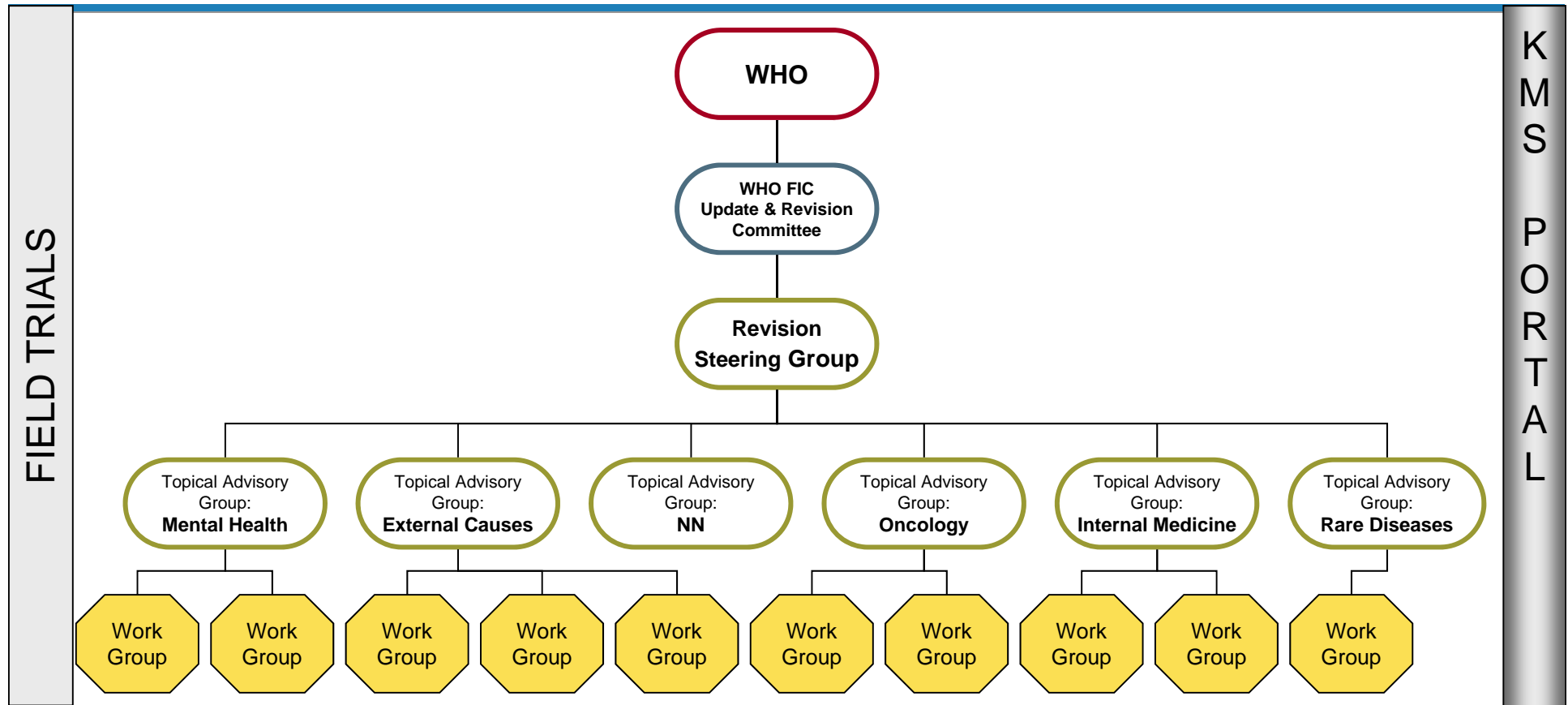


Other fields

- Maternal, neonatal and Child health:
 - Discussions ongoing with WHO Dpts
- Infectious Diseases
 - WHO Dpt, David Heyman (Control of Communicable Diseases Manual)
- Neurology
- Eye, otorhinolaryngoiatrics, skin
- Female/Male Genital Diseases
- Urology
- Congenital malformations, deformations and chromosomal abnormalities



ICD-10 Revision organization structure



ICD-10 Plus

ICD-11 draft

ICD Terminology

Application

ICD-10 +
WEB Application

Hi-Ki
Joint-authoring Tool
WIKI like application

Protégé/OWL
LEXGRID

Key Tasks

- Clinical Modification Owners enter their **CM** version Codes
- TAG and **Workgroups** enter proposals

- TAG Experts for ICD-11
- WHO editors
 - Taxonomic rules
 - Definitions
 - Diagnostic criteria

- Ontology Model
- Linkages between ICD and:
 - SNOMED
 - Other ontology & terminologies
- Clinical interface algorithms

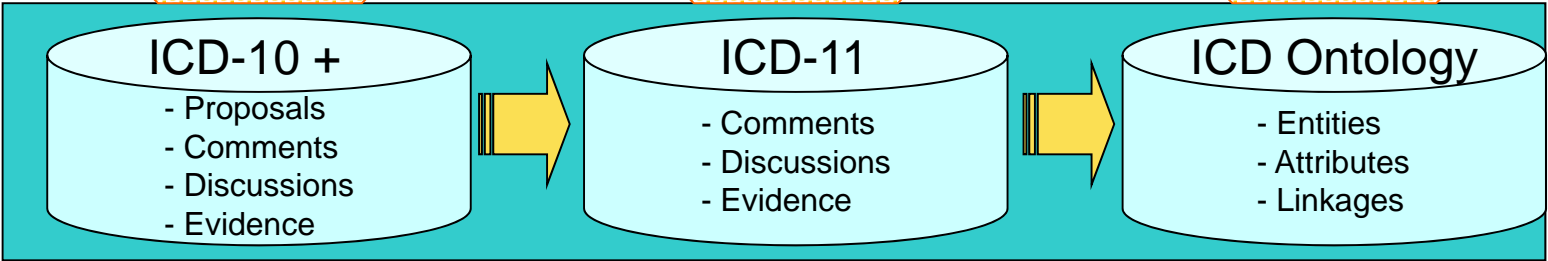
Access

- ANY USER can **POST** proposals or comments.
- ANY USER can **REVIEW** other proposals and discuss.

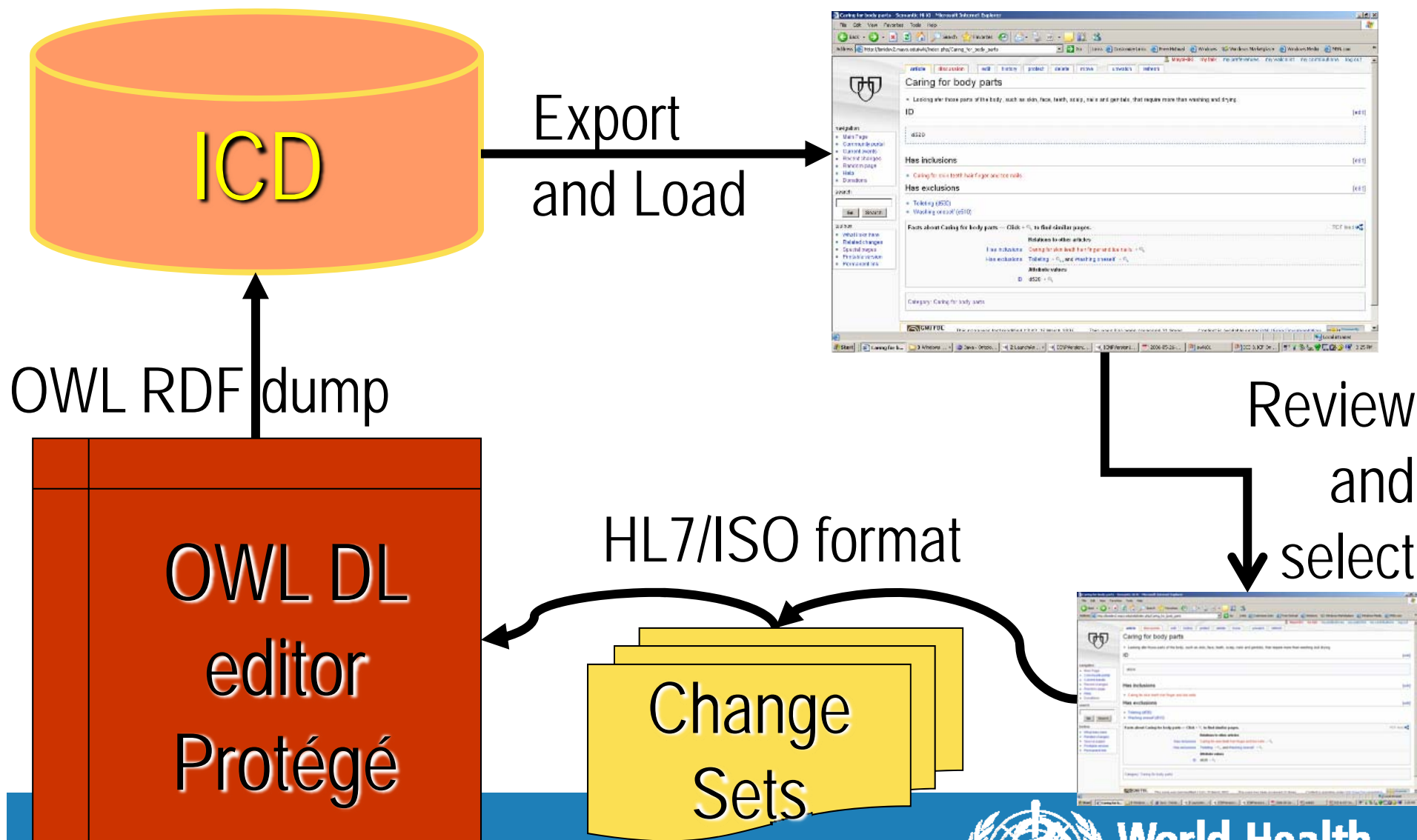
- **ALL USERS** can see drafts and comment.

- **ALL USERS** can see drafts and comment

Technical Layer



Proposed Process draft versioning not shown



Change Proposal Generation

- Implicit creation of change proposal by changing local view
- Can be extracted into machinable format
- Editors and reviewers can see changes in context
 - What would it look like if adopted
- User can write explicit proposal



Hierarchy of Hiki Authority

by ICD Domain

- 0 Revision Steering Committee
- 1 Revision Domain/Topic Working Groups
- 2 Accredited Experts
 - Designated by Working Group Members
- 3 Accredited Persons
 - Designated by Experts
- 4 Registered Interested Persons (Public)



Next Steps

- Depend on the stage of the groups
 - Link existing content to ICD
 - Submit and discuss Proposals for Revision on the platform
 - Structure the content (definitions, diseases) along the criteria mentioned

- Reconvene in Trieste 2007
 - Progress
 - Work programmes until 2010



Breast Cancer

- A primary malignant neoplasm of the breast [breast cancer] (254838004)
 - *is a carcinoma* [*is a malignant proliferation of epithelial cells lining the ducts or lobules*], (68453008)
 - *or a sarcoma* (2424003),
 - *or a lymphoma* (115244002),
- of the breast.
- Metastases of malignant neoplasms of other sites, infiltration of the breast by tumours of adjacent structures and cancer of the skin of the breast are not part of this definition of breast cancer.



Rewriting ICD Using Terminologies

Breast Cancer

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Diabetes mellitus (E10–E14)

Diabetes mellitus in adults is best defined on the basis of the blood glucose concentration measured two hours after a 75g oral glucose challenge, following an overnight fast of 8-14 hours and three days of unrestricted diet. The measurement of the fasting blood glucose alone is less satisfactory but may be used to verify diabetic status in subjects claiming to have pre-existing diabetes.

Random blood glucose measurement has a wide range of uncertainty and is the least recommended method. Subjects taking current, regular insulin or oral hypoglycaemic agents are considered diabetic, regardless of their blood glucose concentration.

Diagnostic values for the oral glucose tolerance test (WHO 1999)

Diabetes mellitus	Glucose concentration, mmol/litre (mg/dl)			
	Whole blood		Plasma	
	Venous	Capillary	Venous	Capillary
Fasting value	≥ 6.1 (≥ 110)	≥ 6.1 (≥ 110)	≥ 7.0 (≥ 126)	≥ 7.0 (≥ 126)
2 hours after glucose load	≥ 10.0 (≥ 180)	≥ 11.1 (≥ 200)	≥ 11.1 (≥ 200)	≥ 12.2 (≥ 200)

Use additional external cause code (*Chapter XX*), if desired, to identify drug, if drug-induced.

The following fourth-character subdivisions are for use with categories E10-E14:



Tentative Timeline

- 2010 : **Alpha version** (ICD 10+ → ICD 11draft)
 - +1 YR : **Commentaries and consultations**
- 2011 : **Beta version & Field Trials Version**
 - 20+2 YR : **Field trials**
- 2013 : **Final version** for public viewing
 - 2014 : **WHA Approval**
- 2015+ **implementation**

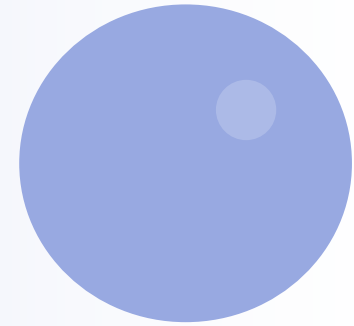


Rewriting ICD Using Terminologies

clinical stage 2 HIV / AIDS

1.	positive HIV antibody test	250514000	165816005
2.	Moderate unexplained weight loss (<10% of presumed or measured body weight)	248349002	
3.	Recurrent respiratory tract infections (Rites, sinusitis, bronchitis, otitis media, pharyngitis)	195708003 (..., 36971009, 32398004, 65363002, 405737000)	
4.	Herpes zoster	4740000	
5.	Angular cheilitis	266429005	
6.	Recurrent oral ulcerations	281775009	
7.	Papular pruritic eruptions	279333002	271757001
8.	Seborrhoeic dermatitis	50563003	
9.	Fungal nail infections of fingers	414941008	91456000
	Mild immunosuppression	255604002	38013005



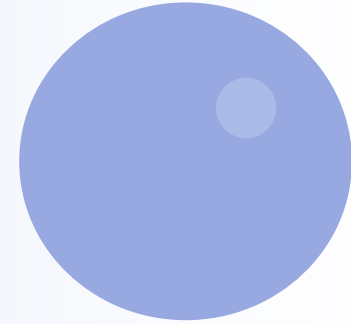


Contribution of Orphanet

To coding and classification of
rare diseases

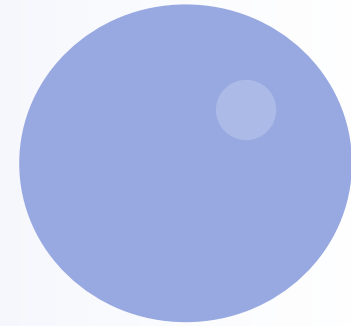
Orphanet

10 years of service



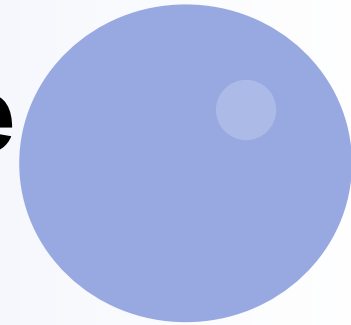
- Established by the French Ministry of Health and by Inserm in 1997
- Funded since 2000 by the European Commission (DG Public Health and Research)
- Budget: 1.5 M€ per year

www.orpha.net



- Orphanet mission
 - Establish a updated list of rare diseases and provide up-to-date information about them, including a directory of services in Europe
- Orphanet achievements
 - Database of 4,300 rare phenotypes
 - Encyclopaedia: 2,023 summary articles and 431 full texts (peer-reviewed)

An Information Service



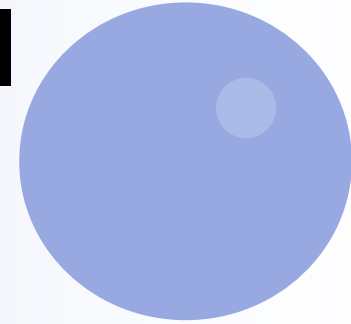
Encyclopaedia

- ▶ Over **4,000** diseases
- ▶ In **6** languages: English, French Spanish, German, Italian, Portuguese
- ▶ Written by experts
- ▶ An international editorial committee
- ▶ Partnerships with journals
- ▶ A free-access electronic journal
Orphanet Journal of Rare Diseases

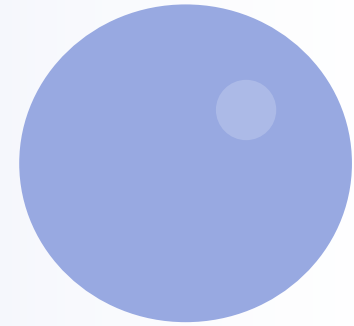
Directory of Resources in Europe

- ▶ **1,078** expert clinics
- ▶ **1,380** clinical laboratories
- ▶ **2,250** research laboratories
- ▶ **450** clinical trials
- ▶ **4,180** research projects
- ▶ **350** registries
- ▶ **85** networks
- ▶ **1,560** advocacy groups
- ▶ **7,230** professionals

Orphanet International Editorial Board

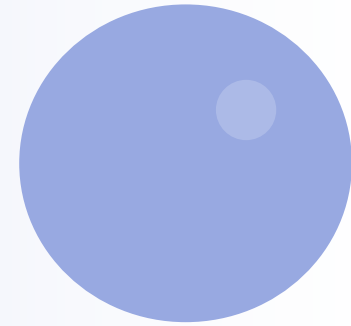


- Established to cover all medical specialties: adulthood and childhood
- Nominations by learned societies
- Terms of reference:
 - Assist the Orphanet team
 - Review editorial content
 - Provide expert advice on coding and classification



Contribution of Orphanet to Classification

Defining Diseases

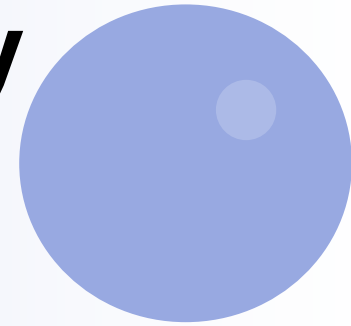


- Example: Ehlers-Danlos disease
 - Dominant form: type 1,2,3,4,7,8, 11
 - X-linked form: type 5, ED with PV heterotopia
 - Recessive form: type 6, 7C, 10, progeroid

1 disease, 3 diseases or 13 diseases ?

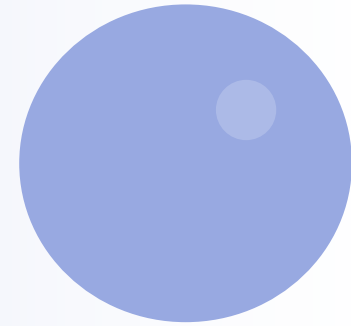
- A disease is a phenotype which is recognisable clinically and which has a unique management approach

Classifications already Identified (1)



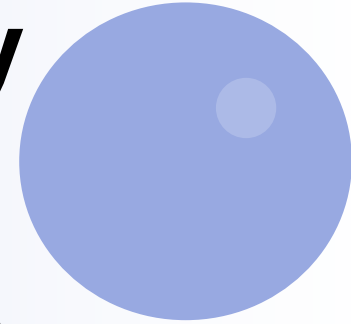
Bone diseases
Genetic Skin Disorders
Classification of diabetes mellitus
Diseases of Skeletal Muscle
Genetically recognized forms of congenital muscular
dystrophies
Distal arthrogryposes
Genetic Dystonia
Ectodermal dysplasia
Ehlers Danlos diseases
Epidermolysis bullosa
Epilepsy
Genetic obesity

Classifications already identified (2)



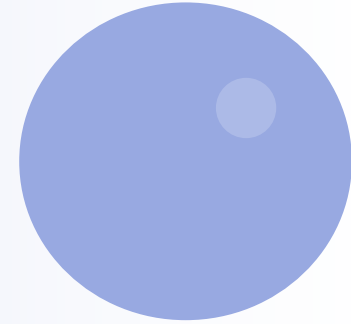
Ichthyoses
Tumours of haematopoietic and lymphoid tissues
Lipodystrophies
Channelopathies
X-linked mental retardation
Male infertility
Mastocytosis
Metabolic Disorders in childhood
Optic Neuropathies
Nuclear encephalopathies
Overgrowth syndromes
Classification of anomalous sexual development

Classifications already identified (3)



Primary immunodeficiency diseases
Autosomal recessive cerebellar ataxias
Dominant spinocerebellar ataxias
Sleep disorders
Charcot-Marie Tooth disease
Syndromic classification of hereditary lymphedema
Congenital macrothrombocytopenias
Neurocutaneous syndromes
Autoimmune uveitis
Congenital heart malformations
Peripheral neuropathies
Vascular malformations
Genetic ophthalmological disorders

Classification of Mastocytosis



Publication: Valent P et al. Diagnostic criteria and classification of mastocytosis: a consensus proposal. *Leuk Res.* 2001 Jul;25(7):603-25

Mastocytosis

Cutaneous mastocytosis

Urticaria Pigmentosa

Typical UP

Plaque-forme

Nodular

Telangiectasia macularis eruptiva perstans

Diffuse cutaneous mastocytosis

Mastocytoma of skin

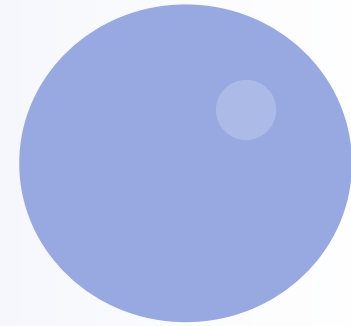
Indolent systemic mastocytosis

Smoldering Systemic Mastocytosis

Isolated bone marrow mastocytosis

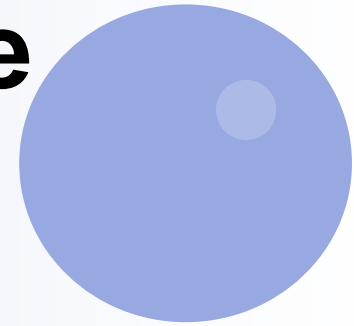
.....

Classification of Channelopathies

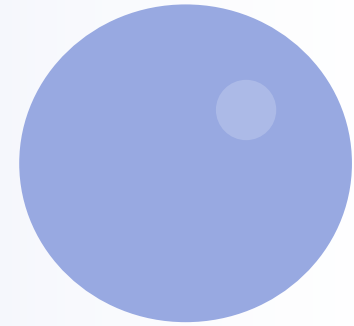


- **Classification 1** : Graves TD, Hanna MG. Neurological channelopathies. *Postgrad Med J*. 2005 Jan;81(951):20-32.
 - Neurological inherited / autoimmunes
- **Classification 2** : Ashcroft FM. From molecule to malady. *Nature*. 2006 Mar 23;440(7083):440-7.
 - Pore-loop / Non-pore-loop channelopathies / cys-loop receptors

Time Table for the Release of Classifications



- All published classifications will be collected by mid 2007
- All classifications will be entered in the Orphanet database by mid-fall 2007
- Classifications will be searchable by January 2008



Contribution of Orphanet to Coding

Indexation of RD in Orphanet

An On-going Process



- ICD-10
 - 324 diseases have a specific code
 - 1,586 have a generic code
- MeSH
 - MeSH terms attributed to 1,149 diseases
- PubMed automatic search tool
 - Available so far for 1,407 diseases

SERVICES FOR :

- Patients
- Professionals
- Support groups
- Industrials
- Public

OrphaNews Europe

- About Orphanet
- About rare diseases
- About orphan drugs
- Support groups

Warning !

- Headquarter
- Contact

Orphanet Reports Series

Print

Search by disease

[Access the alphabetical list of diseases](#)

WHAT'S NEW ?

[Orphanet's 10th birthday
Conference presentation
highlights](#)

[EC Rare Disease webpages](#)

DOCUMENTS

[Centres of Reference for Rare
Diseases in Europe:
State-of-the-art and
Recommendations, Rare Disease
Task Force Dec. 2006](#)

[EC Work Plan 2007 for Rare
Diseases
for DG SANCO's Programme of
Community Action in the field of
Public Health, adopted Feb.07](#)

[French National Plan for Rare
Diseases](#)

[EURORDIS Contribution Paper
on Health services](#)

[European Commission Inventory
of Incentive Measures for Orphan
Medicinal Products](#)

ORPHANET MISSION

ORPHANET is a database dedicated to information on rare diseases and orphan drugs. Access to this database is free of charge.

ORPHANET aims to improve management and treatment of genetic, auto-immune or infectious rare diseases, rare cancers, or not yet classified rare diseases.

ORPHANET offers services adapted to the needs of patients and their families, health professionals and researchers, support groups and industry.

Participating
in clinical trials

Register your
activity

EVENTS

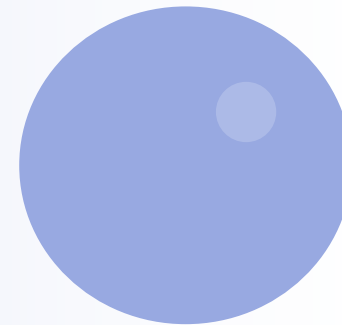
[Calendar of
upcoming events](#)

[39th European Human
Genetics Conference -
Nice, France 16-19
June 2007](#)

8th EPPOSI Workshop
on Orphan Drugs -
Copenhagen 18-19
October 2007 More
details to follow shortly

4th European
Conference on Rare
Diseases (ECRD 2007)
Lisbon, Portugal 27-28
November 2007 More
details to follow shortly

Rare Diseases
Research European
Conference - Brussels,
Belgium 13
September 2007 More
details to follow
shortly



Search by disease

Description of services

Search by clinical signs

Outpatient clinics

Research projects

Clinical trials

Participate in clinical trials

Registries / Observatories

Clinical tests

Accreditations

Support groups





Networks

Laboratories / Departments

Drugs

Professionals

Ask Orphanet

- About Orphanet 
- About rare diseases 
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Orphanet Reports Series 

DISEASE : Prader-Willi syndrome

Orphanet number
ORPHA739

Synonym(s)
Willi-Prader syndrome

ICD Q87.1

Prader-Willi syndrome (PWS) is a chromosomal microdeletion/disomy disorder, sporadic in occurrence, with reported rate of prevalence 1/10,000 to 1/30,000. Severe hypotonia and feeding difficulties in early infancy (age 0 to 2 years), followed by excessive eating, morbid obesity and mental retardation (mostly moderate) in later infancy characterise the syndrome. The cognitive impairment varies in degree and consists in learning disabilities and behavioural troubles. Short stature and lack of pubertal growth spurt are generally present. In 70% of cases PWS arises from deletion of 15q11-13 on chromosome 15. Approximately 25% of patients with PWS have maternal uniparental disomy for chromosome 15; the remainder of patients have a translocation or other structural alteration in chromosome 15. Molecular genetic testing is very important for the initial diagnosis and family genetic counselling, since anticipatory guidance and intervention can significantly influence the outcome. PWS requires symptomatic and preventive treatment that includes management of hypotension and poor feeding initially, followed by management of obesity, scoliosis and behavioural troubles. Growth hormone therapy helps to normalize the height, to increase lean body mass, and is beneficial to weight control. Specific management should be provided for patients with hypothyroidism and hypogonadism. *Authors: Dr. G. Diene and Dr. M. Tauber (January 2005)*.

MIM : [176270](#)

[Scientific publications PubMed](#)

[Clinical signs\(32\)](#)

[Other website\(s\)\(9\)](#)

▶ Outpatient clinic(s)

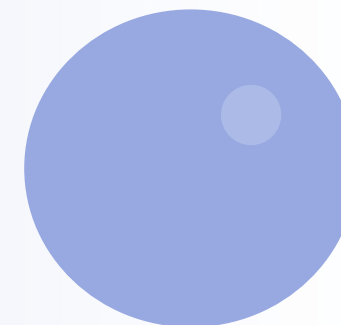
[Genetic counselling clinic](#)

[Genetic obesity clinic](#)

[Prader-Willi clinic](#)

[Rare epilepsies clinic](#)

[Any outpatient endocrinology clinic](#)



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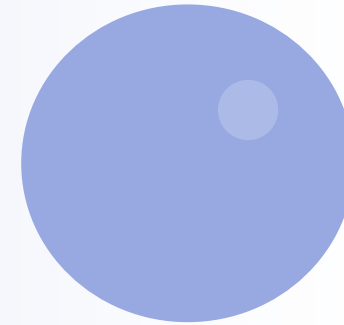
Orphanet Reports Series ◀

Print 

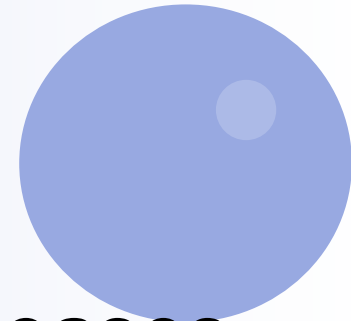
SignList

DISEASE : Prader-Willi syndrome

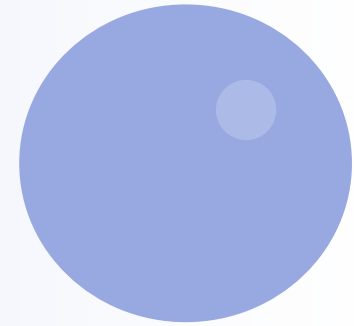
- ▶ ataxia/incoordination (Frequent sign)
- ▶ behaviour disorder/autism (Frequent sign)
- ▶ brachydactyly (Frequent sign)
- ▶ chromosomal anomalie (number) (Frequent sign)
- ▶ clinodactyly of fifth finger (Frequent sign)
- ▶ delayed bone age (Frequent sign)
- ▶ downslanted fissures (Frequent sign)
- ▶ downturned mouth (Frequent sign)
- ▶ ear cartilage deficiency (Frequent sign)
- ▶ enamel anomaly (Frequent sign)
- ▶ generalized obesity (Frequent sign)
- ▶ high vaulted/narrow palate (Frequent sign)
- ▶ hypotonia (Frequent sign)
- ▶ insulino dependent diabetes (Frequent sign)
- ▶ intrauterine growth retardation (Frequent sign)
- ▶ late puberty/hypogonadism (Frequent sign)
- ▶ low hair line (front) (Frequent sign)
- ▶ mental retardation(degree not assessed) (Frequent sign)
- ▶ mental retardation(mild) (Frequent sign)
- ▶ microcephaly (Frequent sign)
- ▶ micropenis/small penis (Frequent sign)
- ▶ renal glomerular defect (Frequent sign)
- ▶ scoliosis (Frequent sign)
- ▶ seizures (any type) (Frequent sign)
- ▶ short foot/brachydactyly of toes (Frequent sign)
- ▶ short stature/dwarfism (Frequent sign)
- ▶ similar process (Frequent sign)



Diseases Features



- Name + synonyms + included diseases
- Summary: clinical definition + incidence/prevalence + etiology + diagnosis + management
- Orpha code + ICD-10 code + MIM code
- MESH terms + link to PubMed
- List of clinical signs and symptoms (controlled vocabulary)



New Version of Orphanet Website in Preparation

Version 4 (V4)

SEARCH
Disease Name → **OK**

- OTHER SEARCH**
- > Orphan Drugs
 - > Support Groups
 - > Research activities
 - > Clinics
 - > Clinical laboratories

Orphanet Today

- Diseases : 4276
- Clinics : 2228
- Laboratories : 3433
- Support groups : 1381
- Professionals : 7919
- Daily visitors : 20000

- RARE DISEASES**
- > Information about a disease
 - > Alphabetical list
 - > Classifications
 - > Diagnostic assistance
 - > Emergency Guidelines
 - > Patient encyclopaedia
 - > Professional encyclopaedia
 - > The Orphanet Journal of Rare Diseases
 - > About rare diseases
 - > Rare diseases policies

- RESOURCES DIRECTORY**
- > Clinics
 - > Reference centres
 - > Clinical laboratories
 - > Research projects
 - > Clinical trials
 - > Registries / Databases
 - > Professionals
 - > Support Groups
 - > Eurogentest
 - > Events / conferences
 - > Register your activity

- ORPHAN DRUGS**
- > Information about an orphan drug
 - > List of orphan drugs
 - > Clinical trials
 - > Register your clinical trial
 - > About orphan drugs

- HEALTH POLICY**
- > About rare diseases
 - > About orphan drugs
 - > The Rare Diseases Task Force

- EDUCATION TOOLS**
- > Training sessions
 - > Glossary
 - > Education Tools

Improve the quality of
medical care for Rare Diseases.
Provide adapted services to
the rare diseases community.



[About Orphanet](#) | [Contact us](#)
[Register your activity](#) | [Make a donation](#)

- Services for professionals**
- > Classifications of rare diseases
 - > Diagnostic assistance
 - > Professional encyclopaedia
 - > The Orphanet Journal of Rare Diseases
 - > Emergency Guidelines
 - > Powerpoint presentations
 - > Newsletters
 - > Register your activity
 - > OrphanXchange

- Services for patients**
- > Information about a disease
 - > Patient encyclopaedia
 - > Clinics
 - > Participate in clinical trials
 - > Clinical studies now recruiting
 - > Support Groups
 - > Contact other patients/families
 - > Training sessions
 - > Glossary
 - > Newsletters

- NEWS / EVENTS**
- 7th EPPOSI Workshop on Partnering for Rare Disease Therapy Development, Madrid 26-27 October 2008
 - NEW: European Commission Inventory of Incentive Measures for Orphan Medicinal Products
 - NEW: EURORDIS Position Paper on Embryonic Stem Cell Research & Therapy

- Services for support groups**
- > Register your support group
 - > Website assistance, creation/hosting
 - > Newsletters

- Services for industry**
- > OrphanXchange
 - > Orphan Drugs
 - > Clinical trials
 - > Newsletters
 - > Register your clinical trial

- MEDIA**
- > Newsletters
 - > Press releases
 - > Events / conferences
 - > About Orphanet



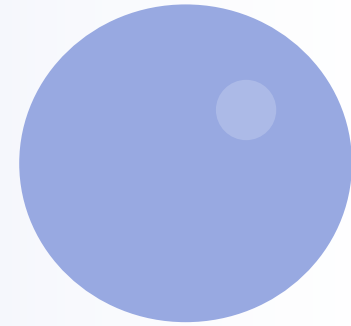
- THE ORPHANET JOURNAL OF RARE DISEASES**
- > Access the journal
 - > Submit a paper

ORPHANET IN COUNTRIES

Austria	Estonia	Italy	Netherlands
Belgium	Finland	Latvia	Norway
Bulgaria	France	Lebanon	Poland
Croatia	Germany	Lithuania	Portugal
Cyprus	Greece	Luxemburg	Romania
Czech Republik	Hungary	Malta	Serbia
Denmark	Ireland	Morocco	Slovakia

Partner access

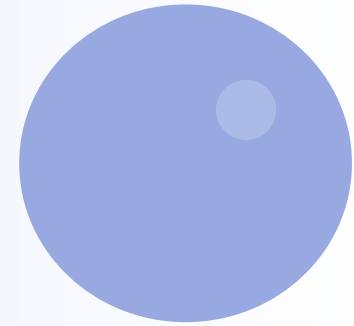
New Features of Diseases



- **Classifications of Rare Diseases / Search Tool**
 - by medical area
 - by mechanism
 - by gene
 - by main clinical expression
 - by aetiology
 - by class of prevalence
 - by inheritance pattern
 - by age at onset

New features for RD in V4

- **Classifications of rare diseases**
 - List of all published classifications
 - Classification of all RD by medical area
 - Visualisation of each classification
 - Possibility to click at any level to have the detailed information
- **Epidemiology of RD**
 - Class of prevalence
 - Age of onset
 - Inheritance



Outcomes of comparisons with other data sets

Outcome typologies



- ICD-10 codes do not match
 - Mistake in one of data sets
 - Different interpretations are possible: needs further examination
- ICD-10 codes match
 - RD is correctly coded in ICD-10 (specific)
 - ICD-10 code is not specific: needs for further examination
 - RD is coded in a wrong ICD-10 category: needs further examination

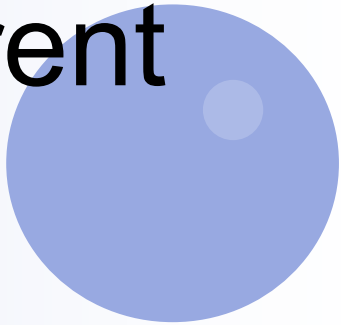
Mismatch due to mistakes

- Multiple endocrine neoplasia
(OMIM 14□31100)
 - UKGTN **D44.8**
 - Pluriglandular involvement .../...Multiple endocrine adenomatosis
 - Orphanet C25.4
 - Malignant neoplasm of endocrine pancreas
C75.0
 - Malignant neoplasm of ...parathyroid gland
C75.1
 - Malignant neoplasm of ... pituitary gland

Mismatch due to mistakes

- Hyperparathyroidism, neonatal severe primary (OMIM 239200)
 - UKGTN E83.5
 - Disorders of calcium metabolism (excludes hyperparathyroidism)
 - Orphanet E21.0
 - Primary hyperparathyroidism

Mismatch due to different interpretations

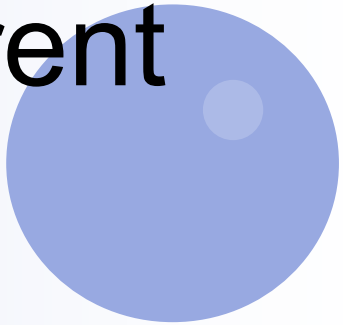


- Barth syndrome (OMIM 302060)
 - UKGTN E88.8
 - Other specified metabolic disorders
 - Orphanet **I42.0**
 - Dilated cardiomyopathy

Mismatch due to different interpretations

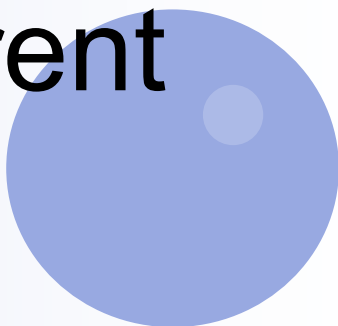
- Cystinosis, nephropatic (OMIM 219800)
 - UKGTN E72.0
 - Disorders of amino-acid transport... Cystinosis
 - N16.3**
 - Renal tubulo-interstitial disorders in metabolic diseases... *Renal tubulo-interstitial disorders in cystinosis*
 - Orphanet E72.0
 - Disorders of amino-acid transport... Cystinosis

Mismatch due to different interpretations



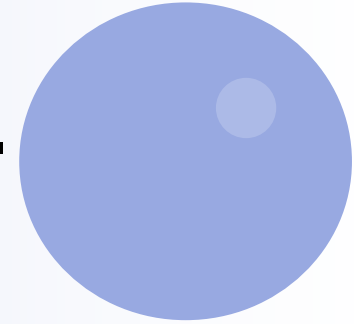
- CADASIL (OMIM 125310)
 - UKGTN 177.8
 - Other specified disorders of arteries and arterioles
 - Orphanet F01.1
 - Multi-infarct dementia (In: vascular dementia)

Mismatch due to different interpretations



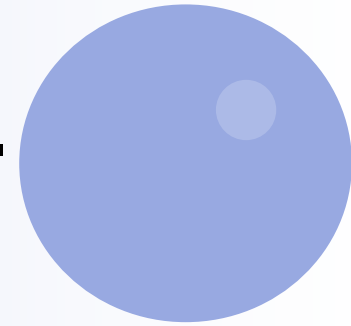
- Norrie disease (OMIM 310600)
 - UKGTN H44.8
 - Other disorders of globe
 - Orphanet Q15.8
 - Other specified congenital malformations of eye

Codes match but...



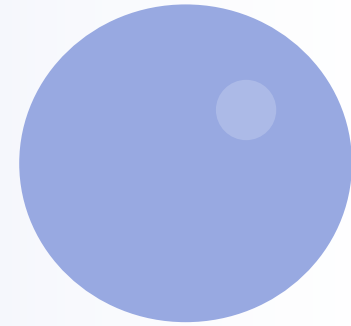
- They are nonspecific
 - Pulmonary lymphangiectasia, congenital
 - CINEAS Q34.8
 - Orphanet Q34.8
 - Other specified congenital malformations of respiratory system
 - There is no code for congenital lung malformations of vascular origin

Codes match but...



- They are specific, but wrong
 - Ehlers-Danlos syndrome type 1
 - CINEAS Q79.6
 - Orphanet Q79.6
 - Ehlers-Danlos syndrome (In: Congenital malformations of the musculoskeletal system, not elsewhere classified)
 - Should be better classified in M00-M99 (Diseases of the musculoskeletal system and connective tissue)

In conclusion



- Cross-mapping data sets allows
 - To identify mistakes and improve coding
 - To identify ICD-10 problems, i.e.
 - Need for categories rearrangement
 - Need for more specific categories, better reflecting homogeneous groups of rare diseases