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Annex 1: STEERING COMMITTEE

PARTNER:	COUNTRY:
Béatrice Gulbis	**Belgium**
Palle Christophersen	**Denmark**
Henri Wajcman	**France**
Max Lakomek	**Germany**
Achile Iolascon	**Italy**
Leticia Ribeiro	**Portugal**
Angel Remacha	**Spain**
Herbert Sandstrom	**Sweden**
Gordon Stewart	**United Kingdom**

Annex 2: MINUTES, SYMPOSIUM 1

Annex 3: DATABASE FIELDS

1) Disease name:
    a. medical terminology
    b. synonyms
    c. abbreviations
    d. keywords (for search possibilities)
2) OMIM link
3) General description of disorder
    a. Professional (symptoms and physical signs)
    b. basic (for MAC) for patient
4) Diagnostic procedure:
   Flowchart
5) Aetiology
6) Heredity
7) Current treatment:
    a. Treatments (where available)
8) Genetic counselling
9) Preventative measures
10) Epidemiology:
    a) Current Situation
    b) Generated from database (dynamic)
        I) Prevalence
        II) Race
        I) Incidence
        II) e-mail address
        III) areas of expertise

11) Specialist centres :

a) country
b) city
c) address
d) telephone number
e) fax number
f) e-mail
g) website
h) services offered: e.g.: phenotype, genotype, etc.
i) specialist registry :
    I) name
    II) country
    III) telephone number
    IV) e-mail address
    V) areas of expertise telephone number

12)Patient organisations

j) Europe
k) Outside Europe

13) Bibliography

a. title
b. authors
l) reference
m) abstract

14)Epidemiology papers

a. title
b. authors
c. reference
d) abstract

15) Projects in progress

16) Last modification date

Annex 4: MINUTES, SYMPOSIUM 2

Annex 5: EXAMPLE TECHNICAL DESCRIPTION

Annex 6: RARE CONGENITAL ANAEMIAS LIST

CONGENITAL RED CELL ENZYMOPATHIES

6-phosphogluconate dehydrogenase deficiency -6PGD-
Adenosine deaminase hyperactivity -ADA-
Adenylate kinase deficiency -AK-
Aldolase deficiency -ALD-
Bisphosphoglycerate mutase-synthetase deficiency -BPGAM-
Enolase deficiency -ENOL-
g-Glutamyl cysteine synthetase deficiency -GCS-
Glucose phosphate isomerase deficiency -GPI-
Glucose-6-phosphate dehydrogenase deficiency -G6PD-
Glutathione peroxidase deficiency -GP-
Glutathione reductase deficiency -GR-
Glutathione synthetase deficiency -GS-
Glyceraldehyde phosphate dehydrogenase deficiency -GAPD-
Hexokinase deficiency -HK-
Lactic dehydrogenase deficiency -LDH-
Phosphofructokinase deficiency -PFK-
Phosphoglycerate kinase deficiency -PGK-
Phosphoglycerate mutase deficiency -PGAM-
Pyrimidine 5' Nucleotidase deficiency -P5'N-
Pyruvate kinase deficiency -PK-
Triose phosphate isomerase deficiency -TPI-

HAEMOGLOBINOPATHIES

alpha thalassaemia -Haemoglobin H Disease-
beta thalassaemia
  Synonym: intermediate thalassaemia
  Synonym: Cooley Disease
Delta Beta thalassaemia -Homozygous-
Haemoglobin C -Harlem-

Haemoglobin C -Homozygous-
Haemoglobin D
Haemoglobin E
Haemoglobin J
Haemoglobin M -associated with haemolysis-
Haemoglobin S
Synonym: Sickle Cell Disease
Heinz bodies anaemias
Hereditary persistance of foetal haemoglobin
Synonym: HPFH
Unstable Haemoglobins

CONGENITAL RED CELL MEMBRANE DEFECTS

Adenosine triphosphatase -ATPase- deficiency
Alpha-beta lipoproteinaemia -ABL-
Atypical hereditary elliptocytosis
Synonym: HE
Cryohydrocytosis -CH-
Hereditary elliptocytosis -HE-
hereditary hydrocytosis -overhydrated form-
Hereditary pseudohyperkaliemia -HPHK-
Hereditary pyropoikilocytosis -HPP-
Hereditary spherocytosis HS (View Details)
Synonym: Minkowski-Chauffard Disease
hereditary xerocytosis -dehydrated form-
Huntington's Chorea
Leach Phenotype
Lecithin cholesterol acyltransferase -LCAT- deficiency
Synonym: Hereditary Lipidic Abnormality of Red Cell Membrane
Synonym: Congenital Target Cell Syndromes
Lutheran null Phenotype
Mc Leod Syndrome
Rh null Syndrome (View Details)
Rhesus-unlinked elliptocytosis
Spherocytic ovalocytosis

CONGENITAL DEFECTS OF ERYTHROPOIESIS

Aceruloplasminaemia (View Details)

Atransferrinaemia -Hypotransferrinaemia-
Autosomal congenital Sideroblastic anaemia -A-CSA-
Blackfan Diamond Syndrome
Congenital defects of Erythropoietin Receptor -Epo-R-
Congenital defects of haptocorrin
Congenital deficency of methylcobalamine
  Synonym: Homocysteinuria
Congenital deficiency of IRP1-IRP2
Congenital deficiency of gut folate conjugase
Congenital deficiency of HFE
  Synonym: Hereditary Haemochromatosis
Congenital deficiency of ileal receptor
  Synonym: Grasbeck Syndrome
  Synonym: Imerslund
Congenital deficiency of Intrinsic Factor
  Synonym: Juvenile Pernicious Anaemia
Congenital deficiency of Ireg 1
Congenital deficiency of Transcobalamin II
  Synonym: Congenital megaloblastic anaemia
Congenital deficiency of transferrin receptor -TfR-
Congenital dyserythropoietic anaemia type I -CDA I- (View Details)
Congenital dyserythropoietic anaemia type II -CDA II- (View Details)
Congenital dyserythropoietic anaemia type III -CDA III- (View Details)
Congenital erythropoietic porphyria -CEP-
Congenital hydrofolate reductase -DHFR-
Congenital hyperhomocysteinaemia
Congenital sideroblastic anaemia with ataxia
Congenital sideroblastic anaemia with erythrocyte dimorphism
Dyskeratosis congenita
Fanconi Anaemia
Hereditary deficeincy of Methyl tetrahydrofolate reductase -MTHFR-
Hereditary folate malabsorption
Hereditary Orotic aciduria
Inherited disorders of cobalamin malabsorption
  Synonym: vitamin B12
  Synonym: Cbl
Iron deficiency anaemia due to congenital intestinal malabsorption
Lesch-Nyhan Syndrome
Pearson syndrome
Purine nucleoside phosphorylase deficiency
  Synonym: PNP
Thiamine-responsive megaloblastic anaemia

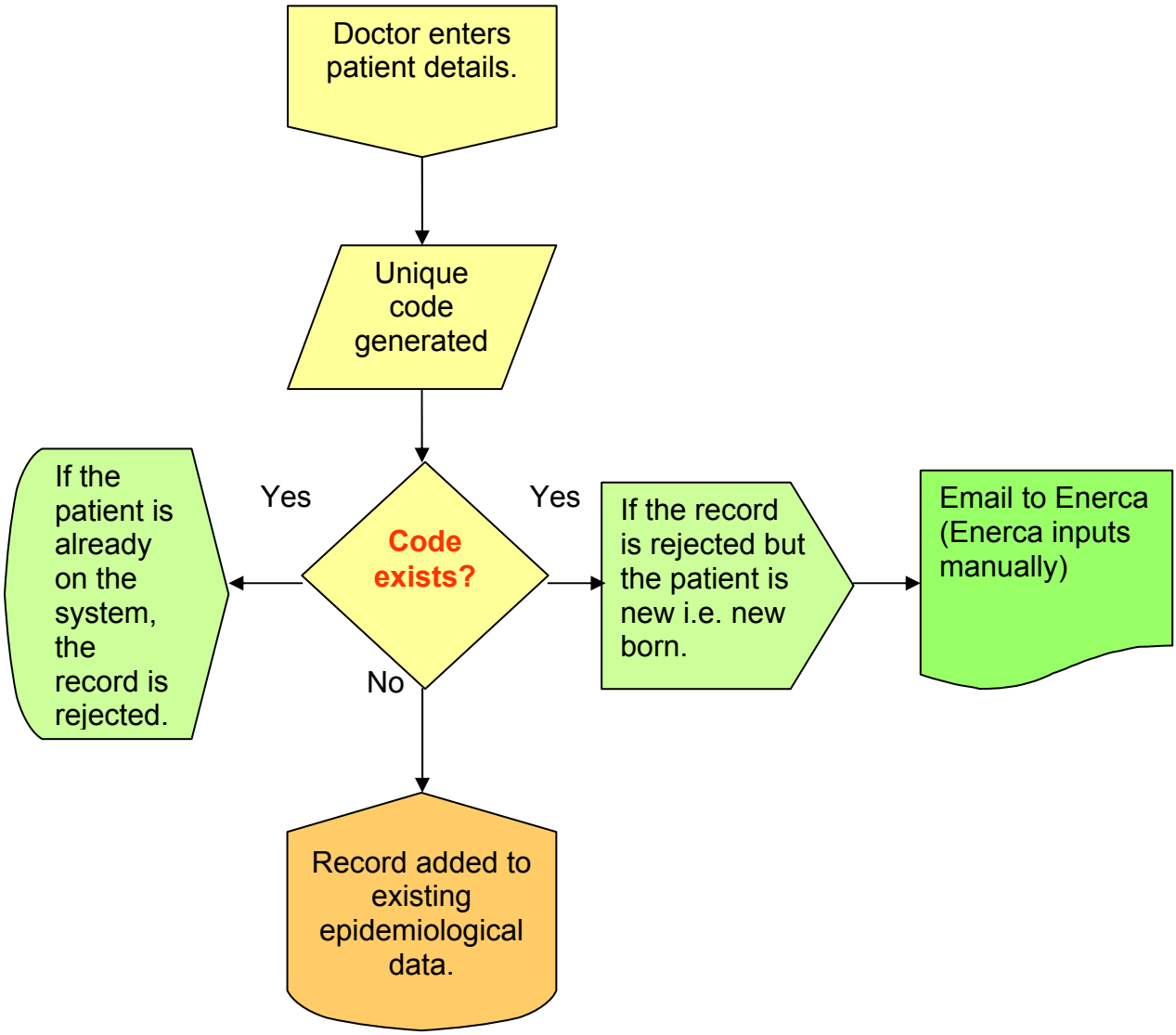
Synonym: TRMA

X-linked congenital sideroblastic anaemia -X-CSA-

Annex 7: WEBSITE OVERVIEW

Annex 8: EXAMPLE MAC

Annex 9: EXPLANATION OF MAC



Annex 10: MINUTES, ITALY TRIP

Annex 11: MINUTES, BELGIUM TRIP

Annex 12: MINUTES, GERMANY TRIP

Annex 13: MINUTES, SWEDEN TRIP

Annex 14: MINUTES, DENMARK TRIP

Annex 15: MINUTES, PORTUGAL TRIP

Annex 16: MINUTES, ENGLAND TRIP

Annex 17: MINUTES, FRANCE TRIP

Annex 18: CONGRESS POSTER

Annex 19: SPONSORS PRESENTATION (In Spanish)

Annex 20: EURORDIS AND ORPHANET COLLABORATION AGREEMENTS

EURORDIS:

Letter of intent - Collaboration between ENERCA and EURORDIS
Draft 1 as of March 10th, 2004

1. EURORDIS and ENERCA will identify in their respective websites the pages where links can be built from one site to the other.

2. ENERCA and EURORDIS representatives will be mutually invited to contribute to conferences concerning actions related to rare congenital anaemias.

3. EURORDIS will include in its newsletter or on its website articles to be provided by ENERCA concerning developments in ENERCA as they arise.

4. ENERCA will provide EURORDIS with epidemiologic advice, on request, as to the collection or interpretation of epidemiologic data on rare diseases.

5. EURORDIS will give ENERCA a yearly report on its evaluation of the ENERCA website from the point of view of patients and patient organisations, suggesting what new information may be added, and how existing information may be clarified for users. This report will be submitted to the ENERCA Steering Committee to decide action, subject to funding.

Paris, March, 2004
(issued in two original copies)

Joan-Lluís Vives Corrons
Project leader
ENERCA

Yann Le Cam
Chief Executive Officer
EURORDIS

ORPHANET:

Letter of intent - Collaboration between ENERCA and Orphanet

Draft 20 April 2004

Further to several contacts, it has been decided to reinforce the collaboration between Orphanet and ENERCA through the search for synergies and complementary approaches in the benefit of users around the ENERCA and Orphanet portals.

1. Content building and links between the two websites

Orphanet and ENERCA agrees to cooperate on content pages of both websites and links to useful databases.

Whenever possible links will be built between the two websites to avoid duplication and increase exchanges.

2. Directory of services

The directory of services (clinical laboratories, specialised clinics, support groups) will be establish in common for the diseases which are covered by ENERCA.

Both teams will transmit to the other the relevant information for updating their website.

More broadly, it is envisaged for both structures to share information on events of interest at European level for publication on both websites as well as other information tools.

The two partners will also invite a member of the other organisation to meetings discussing topics of potential mutual interest.

Paris, 2004

(issued in two original copies)

Joan-LLuis Vives Corrons
Leader of ENERCA team
ENERCA

Ségolène Aymé
Scientific Director
Orphanet

Annex 21: LETTERS FOR PARIS CONGRESS

Annex 22: LETTER ACCEPTING VAT PAYMENT