Hazánk csatlakozása az Orphanet rendszeréhez

Dr. Bencsik Péter
Orphanet, Information Scientist

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Előadás vázlata

- Bemutatkozás
- Az Orphanet bemutatása
- Magyar adatok az Orphaneten
- Tervek

Az Orphanet bemutatása

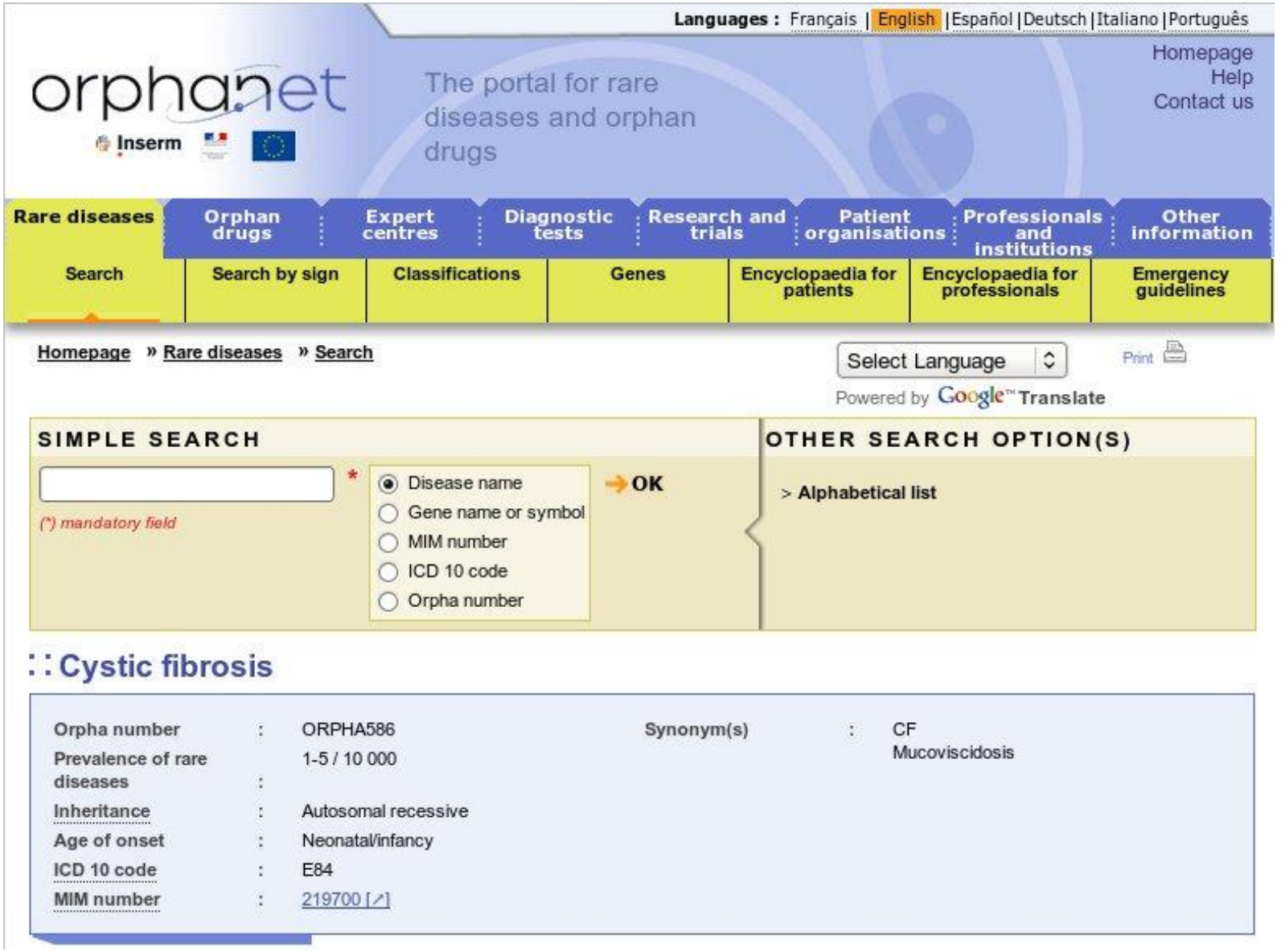
- Felépítés:
  - francia központ: koordináció, infrastruktúra
  - ~40 ország: helyi adatgyűjtés, adatszolgáltatás
- Alapkoncepció: ritka betegség, ritka a szakértő
- Cél: ritka betegségek diagnózisát, kezelését **információval segíteni**
- Referencia oldal a kutatók, orvosok és a betegek számára
- Elérhető: **www.orpha.net**
  - 3-4000 látogató / nap

orphanet

Orphanet szolgáltatások

- **~6000 betegség rendszerezése**: ICD10, OMIM kereszthivatkozás, öröklődés, kapcsolódó gének
- **~2900 betegség enciklopédia** jellegű leírása
- Ún. **árva gyógyszerek** adatbázisa
- **Tagországok szolgáltatásainak adatbázisa**: szakértői központok, kutatások, regiszterek, technikai eszközök, betegszervezetek
- **Diagnózist segítő eszközök**: tünetek alapján lépésről lépésre
- Terápiás **irányelvek**

Enciklopédia példa: Cisztás fibrózis



SUMMARY

Cystic fibrosis (CF) is a genetic disorder characterized by the production of sweat with a high salt content and mucus secretions with an abnormal viscosity. It is the most common genetic disorder among Caucasian children. The incidence varies between populations: the condition is considerably less common in Asian and African populations than in the white populations of Europe and North America, with variation within each country. The exact prevalence in Europe is unknown, but estimates range between 1/8,000 and 1/10,000 individuals. The disease is chronic and generally progressive, with onset usually occurring during early childhood or, occasionally, at birth (meconium ileus). Virtually any internal organ may be involved but the principle manifestations concern the breathing apparatus (chronic bronchitis), pancreas (pancreatic insufficiency, adolescent diabetes and occasionally pancreatitis) and, more rarely, the intestine (stercoral obstruction) or liver (cirrhosis). The most common form of cystic fibrosis is associated with respiratory symptoms, digestive problems (steatorrhea and/or constipation) and staturoponderal growth anomalies. Mortality and morbidity depend on the extent of bronchopulmonary involvement. Male sterility is a constant feature. Late-onset forms, which are usually only mild or monosymptomatic, have also been reported. CF is characterized by alterations in the CFTR protein, which plays a role in the regulation of transmembrane hydroelectrolytic flux. Alterations in the protein lead to changes in the characteristics of exocrine excretions. An absence of functional CFTR in the epithelial cell membrane leads to the production of sweat with a high salt content (associated with a risk of hyponatremic dehydration) and mucus secretions with an abnormal viscosity (leading to stasis, obstruction and bronchial infection). Cystic fibrosis is a monogenic autosomal recessive disease caused by mutations in the CFTR gene (chromosome 7). More than 1250 mutations have been reported. Nearly 70% of all cases are caused by the delta F508 allele, with 30 other mutations accounting for a further 20% of cases. There is no clear correlation between genotype and phenotype. In addition to the allelic heterogeneity and the occurrence of multiple mutations in the same gene, a wide range of other factors may influence the phenotype, including the environment and disease modifying genes. Diagnosis is suspected on the basis of sweat test results (chloride concentration above 60 mmol/L) and is confirmed by identification of a CFTR mutation. Neonatal testing has been widely available since the end of 2002 and leads to diagnosis in 95% of cases. Genetic counseling should be offered to couples carrying heterozygous mutations (identified through the birth of a first child with cystic fibrosis, a family history of the disease or following detection of a heterozygous mutation in an infant screened at birth). Antenatal testing is possible through mutation analysis of chorionic villus samples taken after the eighth week of gestation. Treatment of cystic fibrosis remains purely symptomatic, revolving around bronchial drainage, antibiotics for respiratory infections, pancreatic analysis and administration of vitamins and calorific supplements for digestive and nutritional problems. These cost-effective treatments have significantly improved the prognosis for cystic fibrosis patients: in the 1960's the majority of patients died before 5 years of age, whereas the current average life-span exceeds 35 years and life-expectancy is 40 years. Symptomatic treatment of the disease should improve with the development of etiological treatments with complementary benefits (pharmacological approaches or gene therapy), neonatal testing and multidisciplinary management. *Author: Dr G. Bellon (April 2006)*.

Detailed information

Emergency guidelines	Français [↗] **(2009)**
Clinical practice guidelines	Français [↗] **(2006,pdf)** English [↗] **(2009,pdf)**
Guidance for genetic testing	Français [↗] **(2009,doc)**
Article for general public	Français [↗] **(2006,pdf)**

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Additional information

Further information on this disease

- > Classification(s) (6)
- > Gene(s) (1)
- > Publications in PubMed [↗]
- > Other website(s) (18)

Health care resources for this disease

- > Expert centres (330)
- > Diagnostic tests (368)
- > Patient organisations (64)
- > Orphan drug(s) (48)

Research activities on this disease

- > Research projects (225)
- > Clinical trials (61)
- > Registries/biobanks (37)
- > Licencing offers (10)
- > Networks (23)

Orphanet Reports series

- > Prevalence of rare diseases
- > Orphan drugs in Europe

Getting involved /informed

- > Read the newsletter
- > Read OJRD [↗]
- > Register your activity

Magyarország szerepe, tevékenysége

- **Szakértőket** delegál
  - Nemzetközi Tanácsadó Testületben:
    Dr. Pogány Gábor
- **Adatokat gyűjt**, szolgáltat, frissít
- Kutatási projektekben vesz részt
- Betegek diagnosztizálása, irányítása, kezelése
- Eddigi eredmények

LIST OF PATIENT ORGANISATIONS (BY COUNTRY)

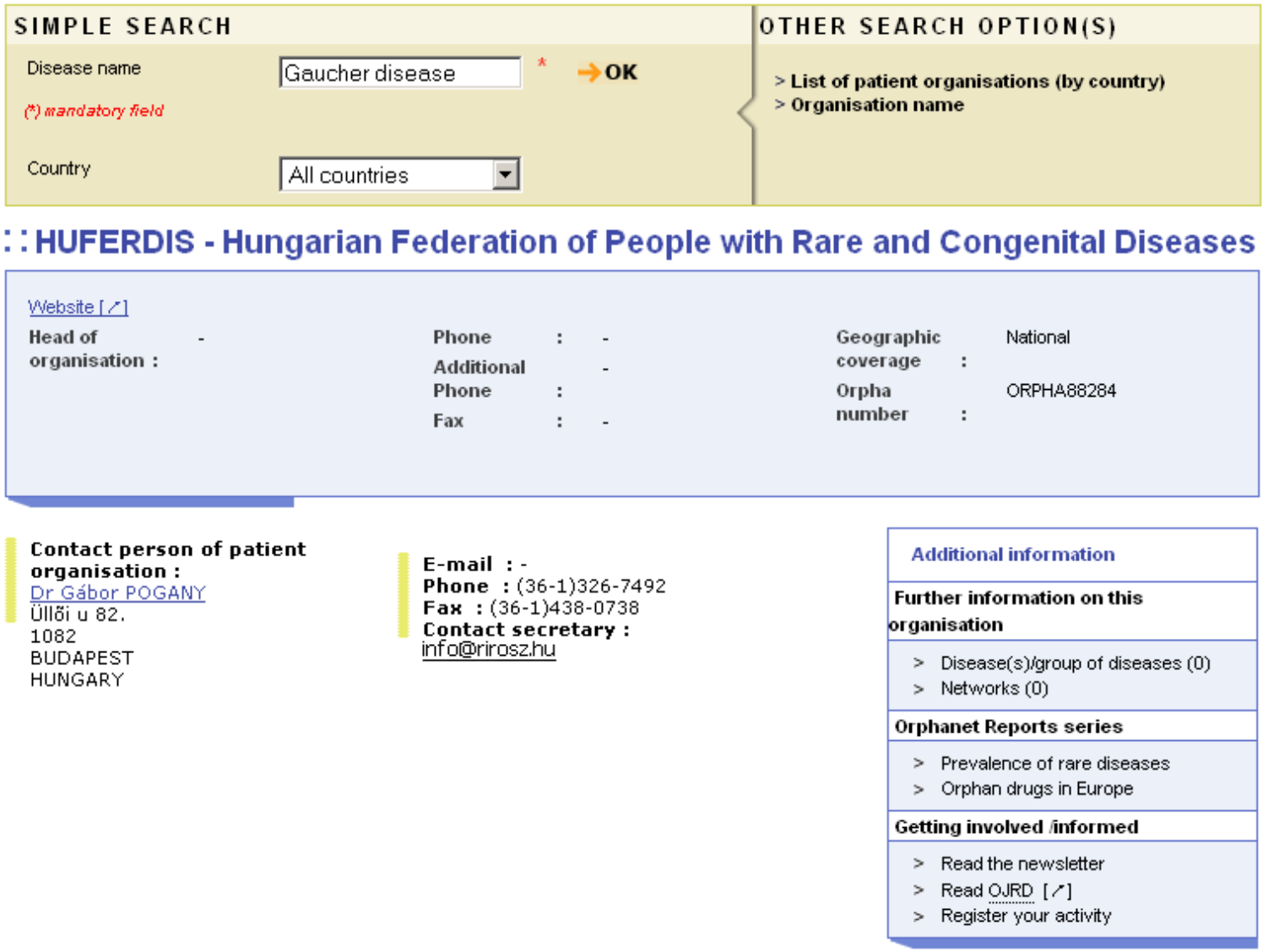
OTHER SEARCH OPTION(S)

> Simple search
> Organisation name

back to list by country

- AOD - Association of Deafblind [↗]
- CDCBT - Cri Du Chat Baráti Társaság [↗]
- CML-GIST - CML és GIST Betegek Egyesület [↗]
- DEBRA Hungary - Országos Epidermolysis Bullosa Centrum - National Centre for Epidermolysis Bullosa [↗]
- DKT - Dávid Kisemberek Társasága [↗]
- Gyermekünk a Holnapunk Kiemelkedöen Közhasznú Alapítvány-Foundation for Children of our tomorrow [↗]
- HUFERDIS - Hungarian Federation of People with Rare and Congenital Diseases [↗]
- HWSA - Hungarian Williams Syndrome Association [↗]
- Magyar Hemofilia Egyesület [↗]
- Magyarorszagi Angio-Odemas Betegek Egyesulete [↗]
- Magyarországi PKU Egyesület [↗]
- MMT - Magyar Mukopoliszaccharidózis Társaság [↗]
- Neurofibromatozisos Betegek es Segitoik Tarsasaga [↗]
- OCFE - Országos Cisztás Fibrózis Egyesület [↗]
- Retina Magyarorszag [↗]

Betegszervezet részletes adatai



SIMPLE SEARCH

Disease name: Gaucher disease * → OK

(*) mandatory field

Country: All countries

OTHER SEARCH OPTION(S)

> List of patient organisations (by country)
> Organisation name

: : HUFERDIS - Hungarian Federation of People with Rare and Congenital Diseases

Website [↗]

Head of organisation : -

Phone : -
Additional Phone : -
Fax : -

Geographic coverage : National
Orpha number : ORPHA88284

Contact person of patient organisation :
Dr Gábor POGANY
Üllői u 82.
1082
BUDAPEST
HUNGARY

E-mail : -
Phone : (36-1)326-7492
Fax : (36-1)438-0738
Contact secretary :
info@rirosz.hu

Additional information

Further information on this organisation

> Disease(s)/group of diseases (0)
> Networks (0)

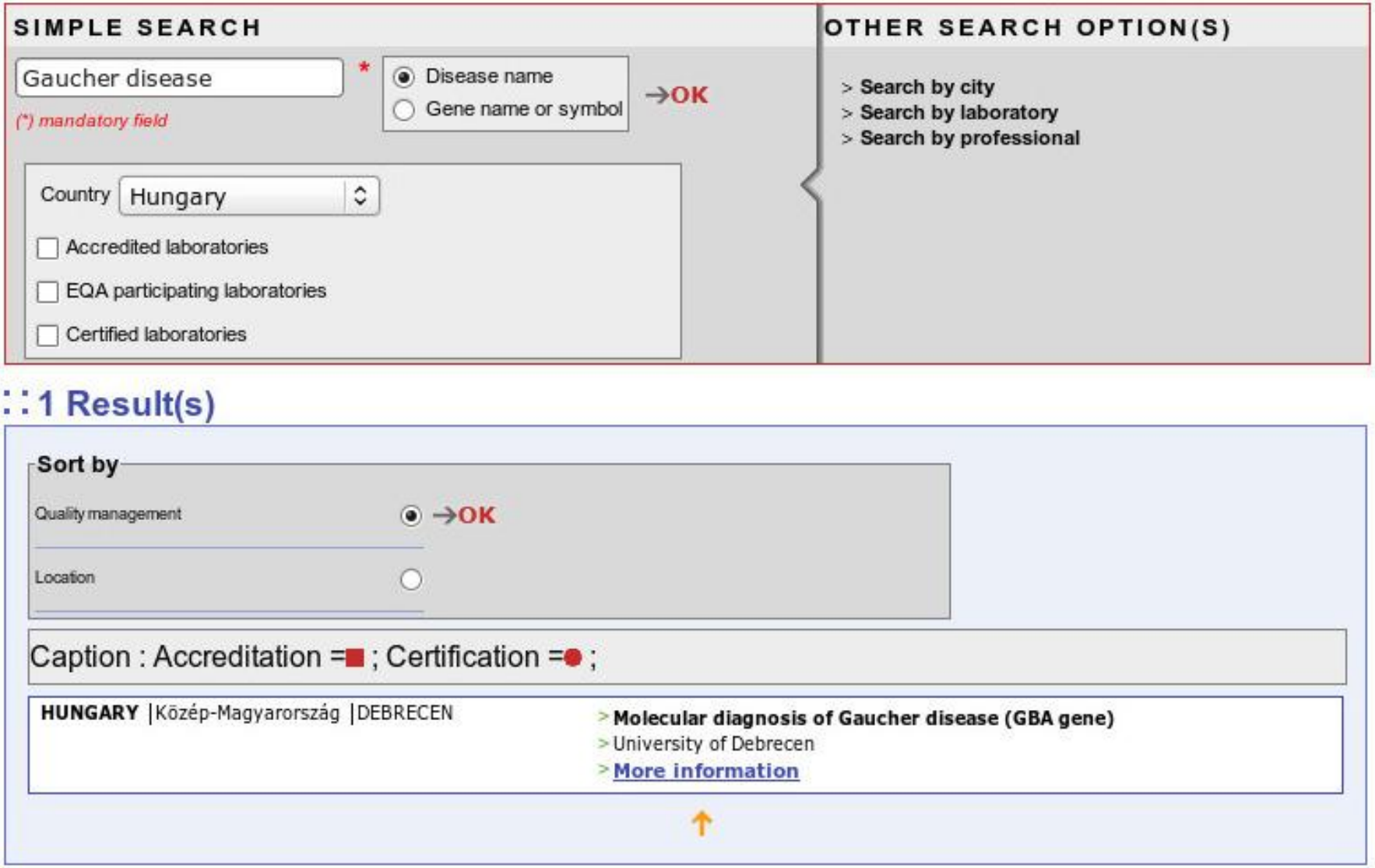
Orphanet Reports series

> Prevalence of rare diseases
> Orphan drugs in Europe

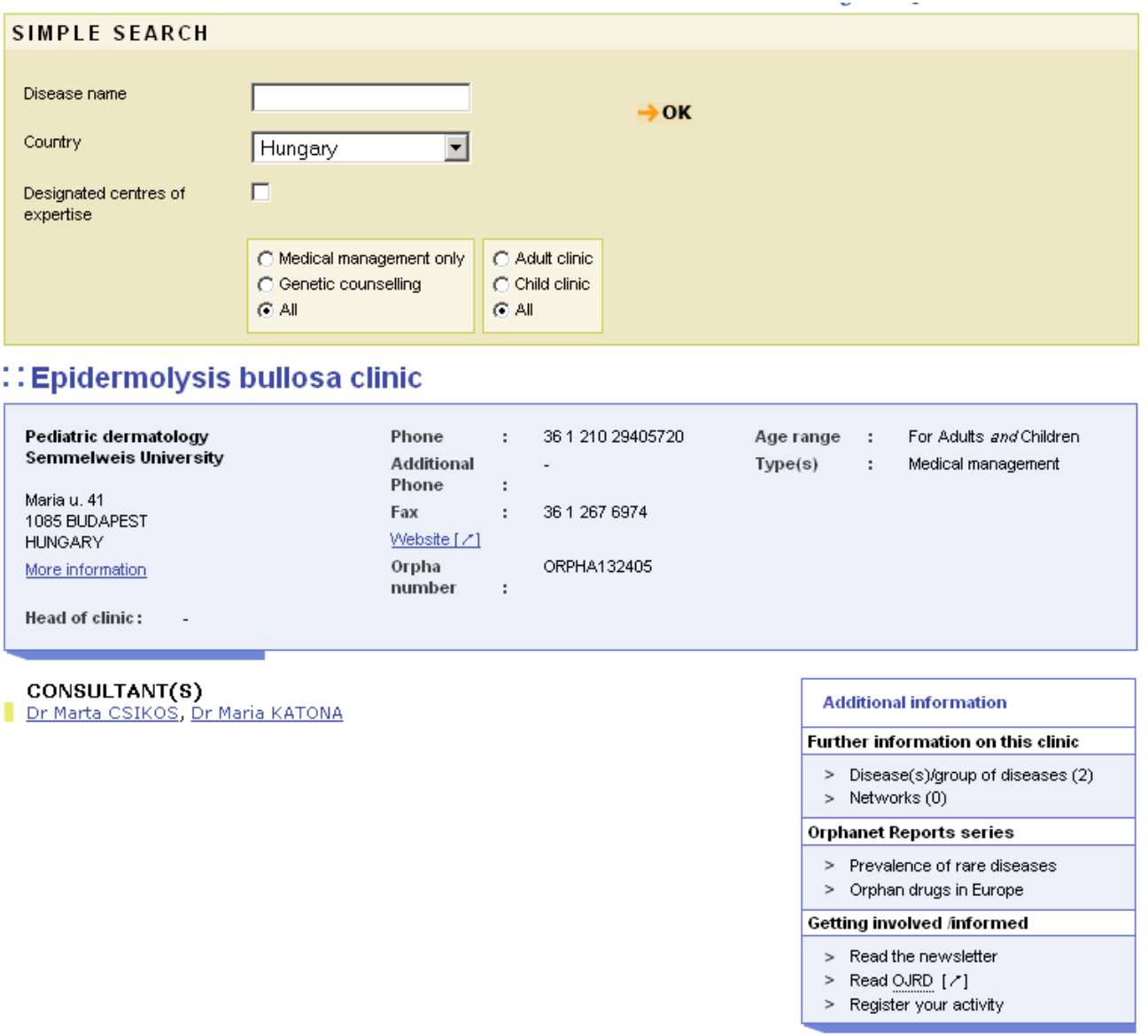
Getting involved /informed

> Read the newsletter
> Read OJRD [↗]
> Register your activity

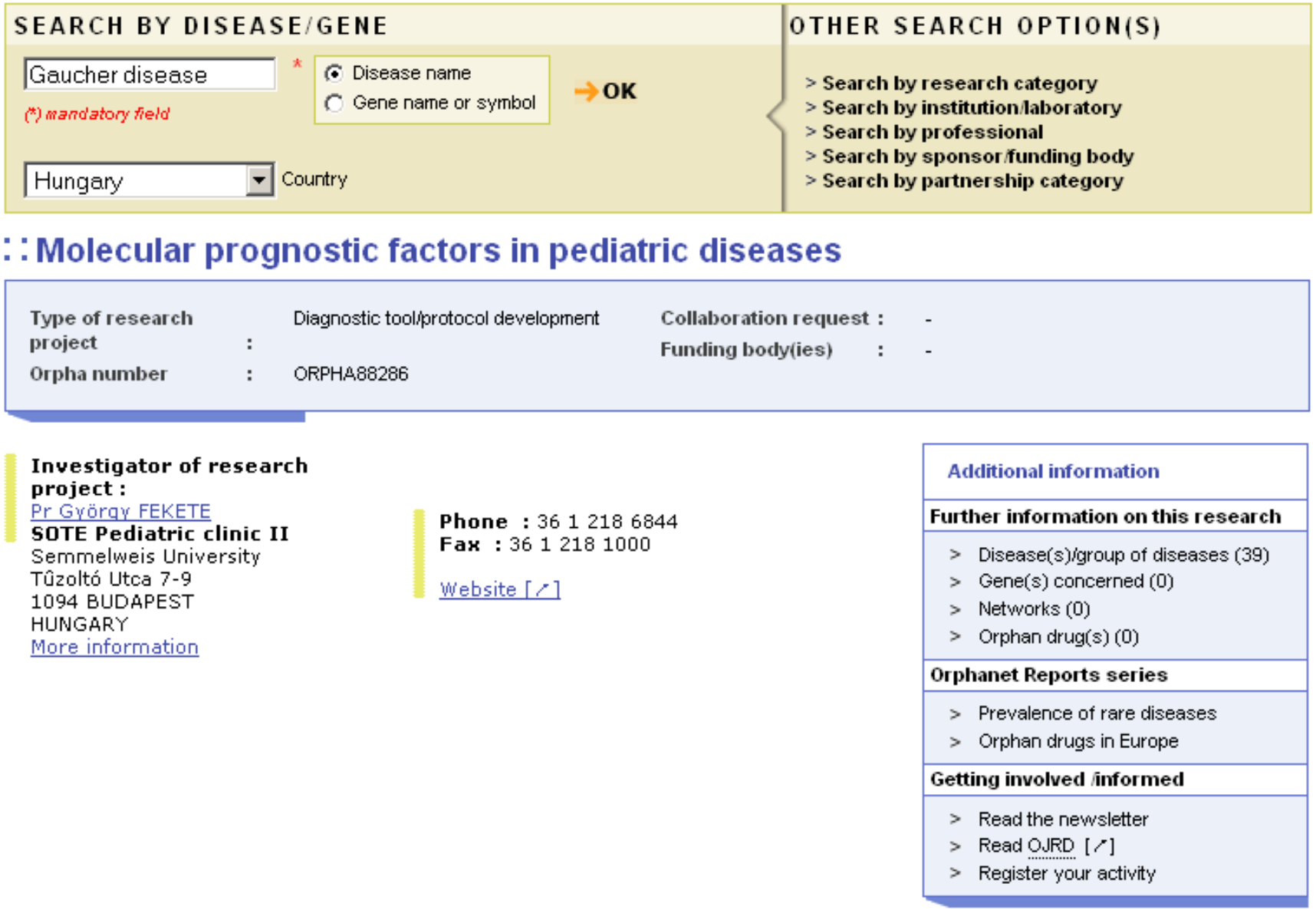
Betegség diagnosztizálásával foglalkozó labor keresése



Szakértői központ megjelenítése

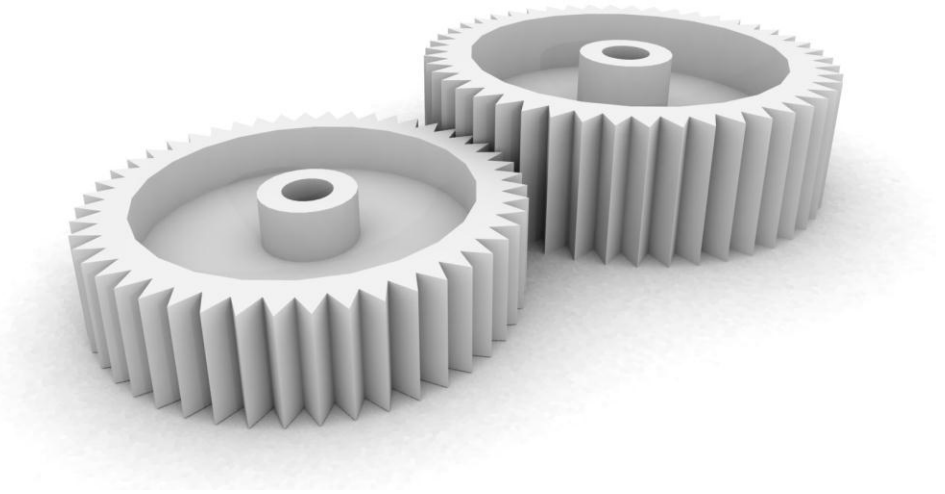


Kutatási projekt megjelenítése



Tervek, teendők

- **Adatgyűjtés**, adatfrissítés folytatása
- **Magyar Orphanet honlap** elindítása
- **Enciklopédia** kiegészítése (Orvosi Hetilap)
- Magyar terápiás **irányelvek** (Napivizit.hu)
- **Betegközösségek** felé kommunikáció (Várószoba.hu)



Összefoglalás

- Az Orphanet **referenciaforrás** a ritka betegségek vonatkozásában
- **Eszközökkel, információval segíti** a betegeket, kutatókat, orvosokat
- Magyarország a 40 közreműködő egyike, eddig is szolgáltattunk adatot, de **részletesebb adatszolgáltatásra, jelenlétre van szükség**

Köszönöm a figyelmet!

Dr. Bencsik Péter

Akadémiai Kiadó, Orvostudományi szegmens

dr.bencsik.peter@akkrt.hu