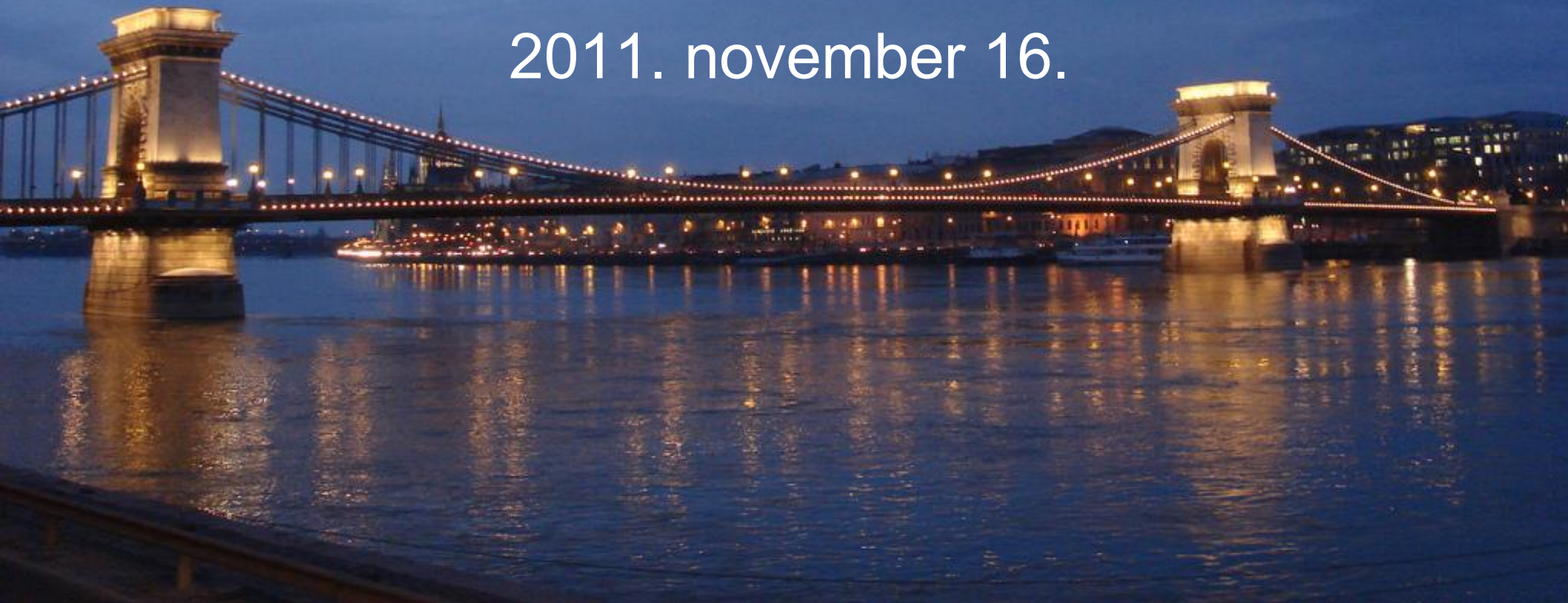


# Hazánk csatlakozása az Orphanet rendszeréhez

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Orphanet, Information Scientist

2011. november 16.



# Előadás vázlata

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



# Az Orphanet bemutatása

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- Felépítés:
  - francia központ: koordináció, infrastruktúra
  - ~40 ország: helyi adatgyűjtés, adatszolgáltatás
- Alapkonceptió: 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

The screenshot displays the Orphanet website interface. At the top, there is a navigation bar with the Orphanet logo and the tagline 'The portal for rare diseases and orphan drugs'. Below this, there are several tabs for navigation: 'Rare diseases', 'Orphan drugs', 'Expert centres', 'Diagnostic tests', 'Research and trials', 'Patient organisations', 'Professionals and institutions', and 'Other information'. A search bar is located in the top right corner. The main content area shows search results for 'Cystic Fibrosis'. It includes a 'SIMPLE SEARCH' section with a search bar and a 'Disease name' dropdown menu. Below this, there is a table with key information about Cystic Fibrosis, such as 'Orphan number', 'Prevalence of rare diseases', 'Inheritance', 'Age of onset', 'ICD-10 code', and 'OMIM number'. A 'SUMMARY' section follows, providing a detailed description of the disease. To the right of the summary, there is an 'Additional information' section with links to 'Further information on this disease', 'Health care resources for this disease', and 'Research activities on this disease'. At the bottom of the page, there is a footer with logos of partner organizations and a language selection menu.

# Enciklopédia példa: Cisztás fibrózis

Languages : Français | **English** | Español | Deutsch | Italiano | Português

**orphane**  
Inserm

The portal for rare diseases and orphan drugs

Homepage  
Help  
Contact us

**Rare diseases** | Orphan drugs | Expert centres | Diagnostic tests | Research and trials | Patient organisations | Professionals and institutions | Other information

Search | Search by sign | Classifications | Genes | Encyclopaedia for patients | Encyclopaedia for professionals | Emergency guidelines

[Homepage](#) » [Rare diseases](#) » [Search](#)

Select Language [Print](#)

Powered by Translate

**SIMPLE SEARCH**

\*

(\*) mandatory field

Disease name → **OK**  
 Gene name or symbol  
 MIM number  
 ICD 10 code  
 Orpha number

**OTHER SEARCH OPTION(S)**

> Alphabetical list

**:: Cystic fibrosis**

Orpha number	: ORPHA586	Synonym(s)	: CF
Prevalence of rare diseases	: 1-5 / 10 000		: Mucoviscidosis
Inheritance	: Autosomal recessive		
Age of onset	: Neonatal/infancy		
ICD 10 code	: E84		
MIM number	: <a href="#">219700</a> [ <a href="#">↗</a> ]		



## 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 staturponderal 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

<b>Emergency guidelines</b>	<a href="#">Français [↗] (2009)</a>
<b>Clinical practice guidelines</b>	<a href="#">Français [↗] (2006,pdf)</a> <a href="#">English [↗] (2009,pdf)</a>
<b>Guidance for genetic testing</b>	<a href="#">Français [↗] (2009,doc)</a>
<b>Article for general public</b>	<a href="#">Français [↗] (2006,pdf)</a>



### 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



The documents contained in this web site are presented for information purposes only. The material is in no way intended to replace professional medical care by a qualified specialist and should not be used as a basis for diagnosis or treatment.

# Magyarország szerepe, tevékenysége

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





Rare diseases	Orphan drugs	Expert centres	Diagnostic tests	Research and trials	<b>Patient organisations</b>	Professionals and institutions	Other information
Search	Services for organisations	Contact other patients	Register / Update your activity	Networks			

Homepage » Patient organisations » Search

Select Language



Powered by Google Translate

**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 Kiseberek Társasága \[↗\]](#)

[Gyermekünk a Holnapunk Kiemelkedően Közhasznú Alapítvány-Foundation for Children of our tomorrow \[↗\]](#)

[HUFERDIS - Hungarian Federation of People with Rare and Congenital Diseases \[↗\]](#)

[HWSA - Hungarian Williams Syndrome Association \[↗\]](#)

[Magyar Hemofília Egyesület \[↗\]](#)

[Magyarországi Angio-Odemas Betegek Egyesulete \[↗\]](#)

[Magyarországi PKU Egyesület \[↗\]](#)

[MMT - Magyar Mukopoliszaccharidózis Társaság \[↗\]](#)

[Neurofibromatozisos Betegek es Segitoik Tarsasaga \[↗\]](#)

[OCFE - Országos Cisztás Fibrózis Egyesület \[↗\]](#)

[Retina Magyarország \[↗\]](#)





# Betegszervezet részletes adatai

SIMPLE SEARCH	OTHER SEARCH OPTION(S)
Disease name <input type="text" value="Gaucher disease"/> * → OK	> List of patient organisations (by country) > Organisation name
(*) mandatory field	
Country <input type="text" value="All countries"/>	

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

<a href="#">Website [↗]</a>			
Head of organisation :	Phone : -	Geographic coverage :	National
	Additional Phone : -	Orpha number :	ORPHA88284
	Fax : -		

### 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](mailto: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

SIMPLE SEARCH	OTHER SEARCH OPTION(S)
<p>Gaucher disease * <i>(*) mandatory field</i></p> <p><input checked="" type="radio"/> Disease name →OK <input type="radio"/> Gene name or symbol</p> <p>Country: Hungary</p> <p><input type="checkbox"/> Accredited laboratories <input type="checkbox"/> EQA participating laboratories <input type="checkbox"/> Certified laboratories</p>	<ul style="list-style-type: none"><li>&gt; Search by city</li><li>&gt; Search by laboratory</li><li>&gt; Search by professional</li></ul>

:: 1 Result(s)

<p>Sort by</p> <p>Quality management <input checked="" type="radio"/> →OK Location <input type="radio"/></p>	
<p>Caption : Accreditation = ■ ; Certification = ● ;</p>	
<p>HUNGARY   Közép-Magyarország   DEBRECEN</p>	<ul style="list-style-type: none"><li>&gt; Molecular diagnosis of Gaucher disease (GBA gene)</li><li>&gt; University of Debrecen</li><li>&gt; <a href="#">More information</a></li></ul>
<p style="text-align: center;">↑</p>	

# Szakértői központ megjelenítése

**SIMPLE SEARCH**

Disease name

Country

Designated centres of expertise

Medical management only  Adult clinic  
 Genetic counselling  Child clinic  
 All  All

→ OK

## :: Epidermolysis bullosa clinic

<b>Pediatric dermatology</b> <b>Semmelweis University</b>	<b>Phone</b> : 36 1 210 29405720	<b>Age range</b> : For Adults <i>and</i> Children
Maria u. 41 1085 BUDAPEST HUNGARY	<b>Additional Phone</b> : -	<b>Type(s)</b> : Medical management
<a href="#">More information</a>	<b>Fax</b> : 36 1 267 6974	
	<a href="#">Website</a> [↗]	
<b>Head of clinic:</b> -	<b>Orpha number</b> : ORPHA132405	

### CONSULTANT(S)

[Dr Marta CSIKOS](#), [Dr Maria KATONA](#)

**Additional information**

**Further information on this clinic**

- > Disease(s)/group of diseases (2)
- > Networks (0)

**Orphanet Reports series**

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

**Getting involved /informed**

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

# Kutatási projekt megjelenítése

SEARCH BY DISEASE/GENE	OTHER SEARCH OPTION(S)
<input type="text" value="Gaucher disease"/> * <i>(*) mandatory field</i>	<ul style="list-style-type: none"><li>&gt; Search by research category</li><li>&gt; Search by institution/laboratory</li><li>&gt; Search by professional</li><li>&gt; Search by sponsor/funding body</li><li>&gt; Search by partnership category</li></ul>
<input type="radio"/> Disease name <input type="radio"/> Gene name or symbol	
<input type="text" value="Hungary"/> Country	

## :: Molecular prognostic factors in pediatric diseases

Type of research project :	Diagnostic tool/protocol development	Collaboration request :	-
Orpha number :	ORPHA88286	Funding body(ies) :	-

### Investigator of research project :

[Pr. György FEKETE](#)  
**SOTE Pediatric clinic II**  
Semmelweis University  
Tűzoltó Utca 7-9  
1094 BUDAPEST  
HUNGARY  
[More information](#)

**Phone** : 36 1 218 6844  
**Fax** : 36 1 218 1000

[Website](#) [↗]

### Additional information

#### Further information on this research

- > Disease(s)/group of diseases (39)
- > Gene(s) concerned (0)
- > Networks (0)
- > Orphan drug(s) (0)

#### Orphanet Reports series

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

#### Getting involved /informed

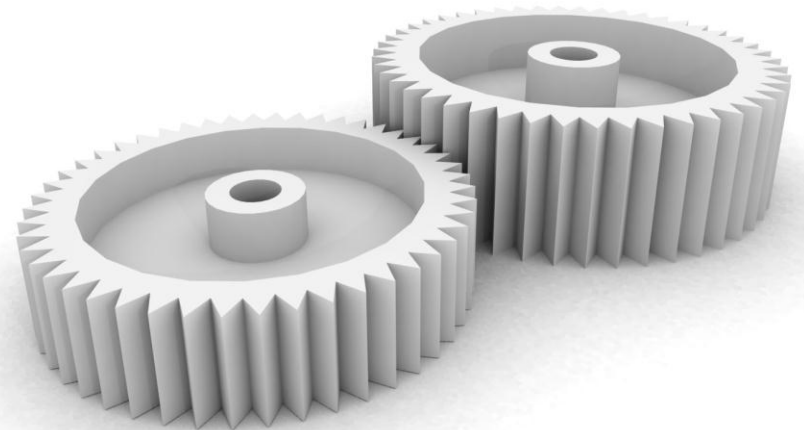
- > Read the newsletter
- > Read OJRD [↗]
- > Register your activity



# Tervek, teendők

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



# Összefoglalás

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



# Köszönöm a figyelmet!



Dr. Bencsik Péter

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