

Eurordis Newsletter November 2007

In this issue:

1. IN BRIEF
2. EDITORIAL
3. NEWS: A one-in-ten-years opportunity for rare diseases
4. SPECIAL REPORT: Commission Conference on Rare Disease Research
5. INSIGHT: United Nations Treaty for people living with disabilities
6. PROFILE: A 30-year battle with muscular dystrophy a.. ORPHAN DRUGS

EDITORIAL

Dear Readers,

Take a break for a moment and let your mind roam free.. The year is 2020. You suffer from a very rare disease and live in Berlin, Rome, Copenhagen, Prague, or Dubrovnik. You are not cured, alas no, but you are alive - with your disease - and you have a place in society as a person living with a rare disease. The last 15 years have seen an improvement of your quality of life and a 10-year increase of your life expectancy, thanks to multidisciplinary expertise- sharing within European reference networks. You benefit from state-of-the-art therapies, thanks to progress made possible by financial contributions from the European Commission and Member States. Workplace discrimination is long forgotten, and you were able to take out a loan to buy your house. Recent information on your disease is available through Orphanet's latest, multilingual version, or through European networks of help lines for rare diseases. You are not alone anymore: you use virtual community tools to communicate on a daily basis with people affected by the same disease in London, Athens or Cracow.

You start thinking about all this progress, those policies shared by Member States and aligned with EU policies, and those outcomes with recognised economic value. You know they came out of national plans for rare diseases and successive EU programmes. And then you remember the Commission Communication on Rare Diseases, initiated in 2007 and adopted in 2008. You remember the momentum of the end of 2007, when every patient group, expert, health industry stakeholder and political decision maker, built the future of rare diseases for the next 15 years.

You too can be part of this movement, by responding to the public consultation on the commission communication on rare diseases! This text will shape your future; it is yours!

Yann Le Cam Chief Executive Officer

NEWS:

A one-in-ten-years opportunity for rare diseases The Public Consultation process for the first Commission Communication on Rare Diseases is about to be launched. The Communication is fantastic good news for the European rare disease community because it will shape the future strategy of Community action in the field of rare diseases for healthcare, research, and therapy development. This article explains what a communication and a recommendation are, then details the content and development process for the current Commission Communication on Rare Diseases, as well as how you can get involved.

Read more >

SPECIAL REPORT: Commission Conference on Rare Disease Research The European Commission organised a conference on rare disease research at the European Parliament on 13 September 2007, entitled "Rare Disease Research: Building on Success". It was another opportunity for the rare disease community to express its needs and to propose a strategic agenda in terms of research. The conference strengthened the

dialogue with the European Commission in the context of the future resource allocation for the 7th Framework Programme for Research and Technological Development.

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INSIGHT: United Nations Treaty for people living with disabilities The United Nations' "Universal Convention on the Rights of Persons with Disabilities", signed at the beginning of 2007, aims to promote, protect and ensure the full and equal enjoyment of all human rights and fundamental freedoms by all persons with disabilities, and to promote respect for their inherent dignity. People living with disabilities represent the world's largest minority. Of these, a large number are also living with a rare disease.

[Read more >](#)

PROFILE: A 30-year battle with muscular dystrophy The Swiss French and Italian-speaking patient organisation for Muscular Dystrophy has fought muscular dystrophy day after day since its inception in 1977. Francine Ludinard looks back over the 30-year struggle: a model of courage and perseverance.

[Read more >](#)

ORPHAN DRUGS

New designations September 07

- Treatment of ornithine-transcarbamylase deficiency Human heterologous liver cells (for infusion)
- Treatment of hereditary factor X deficiency Human coagulation factor X
- Treatment of chronic non-infectious uveitis Cyclo {{{(E,Z)-(2S,3R,4R)-3-hydroxy-4-methyl-2-(methylamino)nona-6,8-dienoyl}-L-2-aminobutyryl-N- methyl-glycyl-N-methyl-L-leucyl-L-valyl-N-methyl-L- leucyl-L-alanyl-D-alanyl-N-methyl-L-leucyl-N-methyl-L- leucyl-N-methyl-L-valyl}}
- Treatment of sarcoidosis Aviptadil
- Treatment of cystic fibrosis Alpha-1 proteinase inhibitor (inhalation use)
- Treatment of cystic fibrosis Alginate oligosaccharide (G-block) fragment
- Treatment of acute myleoid leukaemia 5'-O-(trans-9'-octadecenoyl)-1-β-D-arabinofuranosyl cytosine
- Treatment of hepatocellular carcinoma 4-Amino-1-[5-O-[(2R,4S)-2-oxido-4-(4-pyridinyl)-1,3,2-dioxaphosphorinan-2-yl]-β-D-arabinofuranosyl]-2(1H)- pyrimidinone
- Treatment of Hodgkin lymphoma N-(2-amino-phenyl)-4-[(4-pyridin-3-yl-pyrimidin-2- ylamino)-methyl] benzamide
- Treatment of tuberculosis N-adamantanyl-N'-geranyl-ethylenediamine
- Treatment of renal cell carcinoma Naptumomab estafenatox
- Treatment of cutaneous forms of lupus erythematosus R-salbutamol sulphate
- Treatment of hepatocellular carcinoma Sulfonated monophosphorylated mannose oligosaccharide
- New Marketing Authorisations

- Yondelis [trabectedin] Pharma Mar S.A., Spain
- Yondelis is used to treat patients with advanced soft tissue sarcoma, a type of cancer that develops from the soft, supporting tissues of the body. It is used when treatment with anthracyclines and ifosfamide (other anticancer medicines) have stopped working, or in patients who cannot be given these medicines. Because the number of patients with soft tissue sarcoma is low, the disease is considered 'rare', and Yondelis was designated an 'orphan medicine' (a medicine used in rare diseases) on 30 May 2001. The medicine can only be obtained with a prescription.
- Gliolan [5-aminolevulinic acid hydrochloride] Medac GmbH, Germany
- Gliolan is used in adult patients with malignant glioma (a type of brain tumour). Gliolan helps surgeons to see the tumour more clearly during an operation to remove it from the brain. Because the number of patients with malignant glioma is low, the disease is considered 'rare', and Gliolan was designated an 'orphan medicine' (a medicine used in rare diseases) on 13 November 2002. The medicine can only be obtained with a prescription.
- Increlex [mecasermin] Tercica Europe Limited, Ireland
- INCRELEX is used for the long-term treatment of growth failure in children and adolescents who have severe primary deficiency (low blood levels) of a hormone, insulin-like growth factor-1 (IGF-1), which is required for growth. Patients with this disease produce growth hormone (GH) but their body does not respond to it, and they are short for their age. It is a 'primary' deficiency, meaning that there is no other identified cause for the low levels of IGF-1, such as malnutrition, low levels of thyroid hormone or the use of medicines called steroids to reduce or prevent inflammation. For more information, see the Summary of Product Characteristics (also part of the EPAR). Because the number of patients with primary IGF-1 deficiency is low, the disease is considered 'rare', and INCRELEX was designated an 'orphan medicine' (a medicine used in rare diseases) on 22 May 2006. The medicine can only be obtained with a prescription.
- Atriance [nelarabine] Glaxo Group Limited, United Kingdom
- Atriance is used to treat patients with T-cell acute lymphoblastic leukaemia (T-ALL) or T-cell lymphoblastic lymphoma (T-LBL). These are types of cancer where T-lymphoblasts (a type of immature white blood cell) multiply too quickly. In T-ALL the abnormal cells are mainly in the blood and bone marrow, and in T-LBL they are mainly in the lymphatic system (lymph nodes or thymus gland). Atriance is used when patients have failed to respond to, or have stopped responding to at least two types of chemotherapy. Because the number of patients with these diseases is low, the diseases are considered 'rare', and Atriance was designated an 'orphan medicine' (a medicine used in rare diseases) on 16 June 2005. The medicine can only be obtained with a prescription.

News in brief

- Eurordis mailing lists: an update The Eurordis mailing list service enables people affected by rare diseases in Europe to exchange information and break out of isolation. In 2007, 8 new lists (of a total of 17) have been created. To date, there have been more than 172 000 email exchanges among 476 subscribers. Subscription to the mailing lists is free!
- The Eurordis 2008 Photo Contest is now officially open

'Living with a Rare Disease' The contest is open to anyone having an interest in rare diseases in Europe, whether members of Eurordis or not. You have until the 15th of January 2008 to send your most beautiful, unusual, or artistic photos. Only digital photos are accepted. You can submit up to five photos.

email: eurordis@eurordis.org phone: +33 1 56 53 52 10 web: <http://www.eurordis.org> </FON

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Editorial Team : Yann Le Cam, Jérôme Parisse-Brassens, Julia Fitzgerald, Nathacha Appanah (Writer), Anja Helm, François Houyez

Translation Team : Conchi Casas Jorde (Spanish), Ana Cláudia Jorge and Victor Ferreira (Portuguese), Roberta Ruotolo (Italian), Trado Verso (French), Ulrich Langenbeck (German)

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IN BRIEF Events

Irish Medicines Board (IMB) Information Conference "Medicines for Rare Diseases: An Opportunity for Patients, Science and Industry" 9 November 2007 Dublin, Ireland

Euro-Ataxia 2007 9-10 November 2007 Paris, France

Greek Alliance of Rare Diseases Conference "Orphan Drugs: Access of the Patients with Rare Diseases" 13 November 2007 Athens, Greece

1st Pan-European Workshop on Thalassaemia 26 November 2007 Lisbon, Portugal

4th European Conference on Rare Diseases (ECRD 2007) 27-28 November 2007 Lisbon, Portugal

IAPO Global Patients Congress 2008 20-22 February 2008 Budapest, Hungary

Rare Disease Congress in Latin America - ER2008LA 27-29 March 2008 Buenos Aires, Argentina