



HAL
open science

<http://www.euro-wabb.org>: an EU Register for Alstrom, Bardet Biedl and other rare syndromes

Timothy Barrett, Amy Farmer, Ségolène Aymé, Pietro Maffei, S Mccafferty, Wojciech Mlynarski, Virginia Nunes, Véronique Paquis, Kay Parkinson, Jacques Rohayem, et al.

► To cite this version:

Timothy Barrett, Amy Farmer, Ségolène Aymé, Pietro Maffei, S Mccafferty, et al.. <http://www.euro-wabb.org>: an EU Register for Alstrom, Bardet Biedl and other rare syndromes. First International Cilia in Development and Disease Scientific Conference, May 2012, Londres, United Kingdom. pp.P2. inserm-00752964

HAL Id: inserm-00752964

<https://inserm.hal.science/inserm-00752964>

Submitted on 16 Nov 2012

HAL is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

<http://www.euro-wabb.org>: an EU Register for Alstrom, Bardet Biedl and other rare syndromes

T Barrett^{1*}, A Farmer², S Aymé³, P Maffei⁴, S McCafferty⁵, W Mlynarski⁶, V Nunes⁷, V Paquis⁸, K Parkinson⁹, J Rohayem¹⁰, R Sinnott¹¹, V Tillmann¹², L Tranebjaerg¹³

From First International Cilia in Development and Disease Scientific Conference (2012)
London, UK. 16-18 May 2012

Alstrom syndrome (infancy onset obesity, cardiomyopathy, retinal dystrophy and renal complications) and Bardet Biedl syndrome (polydactyly, infancy onset obesity, retinal dystrophy and learning difficulties) are uncommon (less than 1:100,000), linked by obesity, vision loss and deafness, frequently develop diabetes mellitus by adulthood, and share ciliopathy as the underlying pathology. Delayed diagnosis is common; treatable complications are often missed; and access to molecular genetic testing is unequal between European citizens. There are as yet no orphan drug treatments available, and no access to well characterized cohorts of patients to undertake research. We aimed to establish a European Registry to address these issues. We agreed a common dataset of clinical, investigation and molecular diagnostic data to distinguish between these and other rare syndromes. We wrote an ethics submission template for national approvals, to include consent to link national and international registries. We designed a web based registry with built in security for data confidentiality, anonymised data collection, and facility for patients to self register. Finally we designed a website for dissemination of information to health professionals and families. The core dataset includes 44 data fields which define and separate the syndromes; the extended dataset comprises 370 fields of detailed phenotyping information. We currently have ethics approval in 6 EU states, and the first 40 patients consented, mainly from Italy and UK. This EU Registry will aid the development of national management guidelines and education material for health professionals; improve patient services, raise awareness, and allow recruitment into multi-national clinical trials.

<http://www.euro-wabb.org>

Author details

¹University of Birmingham, UK. ²Birmingham Children's Hospital, UK. ³INSERM, France. ⁴University of Padova, Italy. ⁵University of Glasgow, UK. ⁶University of Lodz, Poland. ⁷IDIBELL, Spain. ⁸Centre Nationale de la Recherche Scientifique, France. ⁹Alstrom Syndrome UK. ¹⁰University of Dresden, Germany. ¹¹University of Melbourne, Australia. ¹²University of Tartu, Estonia. ¹³University of Copenhagen, Denmark.

Published: 16 November 2012

doi:10.1186/2046-2530-1-S1-P2

Cite this article as: Barrett *et al.*: <http://www.euro-wabb.org>: an EU Register for Alstrom, Bardet Biedl and other rare syndromes. *Cilia* 2012 1(Suppl 1):P2.

Submit your next manuscript to BioMed Central and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at
www.biomedcentral.com/submit



¹University of Birmingham, UK
Full list of author information is available at the end of the article