

EURO-WABB

An EU Registry for Wolfram,
Alström, Bardet-Biedl and other rare
Syndromes



www.euro-wabb.org



Increasing knowledge,
raising awareness
and improving the lives of
people and families affected
by WABB syndromes

The Euro-WABB Project has received funding from the European Union in the framework of
the Health Programme



Institute
thématiques

Inserm

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de la santé et de la recherche médicale



Alström Syndrome UK
Support Group



UNIWERSYTET
MEDYCZNY
W ŁODZI



University
of Glasgow

National e-Science
Centre

IDIBELL
Institut d'Investigació Biomèdica de Bellaterra

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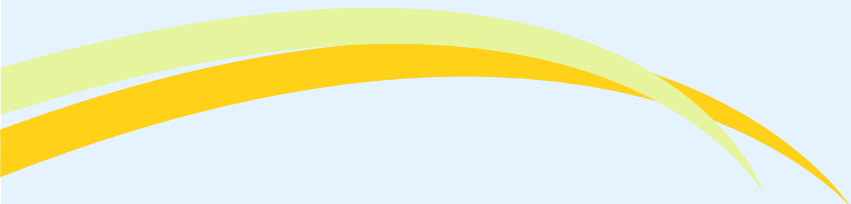
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INTRODUCTION

Recent years have seen increasing interest and resources applied to understand rare diseases. Wolfram, Alström, Bardet Biedl and other syndromes are ultra rare genetic diseases sharing the characteristics of loss of vision, loss of hearing, and a predisposition to diabetes mellitus. Other complications may involve almost any organ system in the body. The improving knowledge of basic disease mechanisms has raised hopes for new treatments to slow or stop the progress of these diseases. However, translational research towards new treatments has faced several barriers.

For patients, promising research results have still not been translated into the treatments they hope for, while lack of standardized care guidelines prevents many from receiving optimal care. Sadly, many families still experience delayed diagnosis and lack of access to diagnostic testing.

For the biomedical industry, identifying investigators and sites with the relevant expertise, and accessing the appropriate patient cohorts for clinical trials, is challenging.

For clinicians and researchers, lack of support tools such as validated clinical outcome measures or standard operating procedures for research protocols has held back development of treatments.

EURO-WABB brings together stakeholders in the community, and provides an infrastructure to accelerate research and therapy development, increasing collaboration, improving patient care and providing a platform to support future international clinical trials .



EURO-WABB was initiated on 1st January 2011 with co-funding from the EU within The Health Programme Framework. The general objective of the project is to support efficient diagnosis, treatment, and research for Wolfram, Alström, Bardet-Biedl (WABB) and other rare syndromes.

The project is managed by a collaboration of scientists, clinicians, and patient groups.

Professor Timothy Barrett
EURO-WABB Project Coordinator



EURO-WABB
partners meeting,
Paris, 2010



CREATING PARTNERSHIPS

Partnering with patient advocacy groups

Close relationships with the major advocacy groups in the field are key to the project's success. The initial European Union funding for the project was advocated by The Association Syndrome de Wolfram, with the support of The Association Française contre les Myopathies (AFM), and the European Organisation for Rare Diseases (EURORDIS). Researchers and clinicians within EURO-WABB benefit from the knowledge and experience that patient groups can provide.

Association Syndrome de
Wolfram:
www.association-du-syndrome-de-wolfram.org/



This association was founded in 2008 by Nolwen LE FLOCH, a teacher, after one of her children was diagnosed with Wolfram Syndrome. The first goal of the association was to initiate research programs on Wolfram syndrome. The association organized the first international workshop on Wolfram syndrome in Paris in 2009. It was decided that a European Registry was absolutely necessary and with a consortium of organizations, EURO-WABB was born. The Association has organized four international workshops of researchers and clinicians; and initiated a research program on gene therapy for the eye, led by a French team, bringing together international collaborators Prof Tanizawa (from Japan), and Profs S Koks and V Tillmann (from Estonia) who agreed to share their resources. The first trials in mice started in April 2013. (<http://loeildelynxmonblog.wordpress.com/2012/07/25/cecile-delette-rechercheuse-inserm-sattaque-au-syndrome-de-wolfram/>). The French Association has also funded a study by an English team with Professor Tim Barrett entitled "Developing personalized therapies for children and adults with Wolfram syndrome", to address the neurological complications; and a French team led by Prof Karsenty (Marseille) has made a preliminary study about urological affection, disseminated it to all the French families and foreign families (http://www.urofrance.org/science-et-recherche/base-bibliographique/resultats/affichage/standard/tri/chronologique-inverse.html?tx_axdocdb_pi1%5Bsword%5D=karsenty)

The Association du Syndrome de Wolfram also creates information leaflets, flyers and annual newsletters. Each year, the French families are invited to the annual meeting of the association. They can meet clinicians and researchers in the morning to ask all questions they have and in the afternoon can discuss with other families to share advice, experience and to share their contact details to communicate throughout the year. After 4 years, the association counts nearly 40 patients in France who communicate regularly. The Association du syndrome de Wolfram plans to organize a multidisciplinary clinic in France in 2014 and the action of the association gave a dynamic who initiated multidisciplinary consultation in the USA and Spain .

CREATING PARTNERSHIPS cont.

Alström syndrome UK:

www.alstrom.org.uk

Alström Syndrome UK was founded in 1998 by Kay Parkinson, after her two children were diagnosed aged 15 and 18 with Alström Syndrome. Initially 7 families were known in the UK and through attendance at the yearly Family Conference, ASUK was able to instigate and develop patient led multi-disciplinary clinics which commenced in 2006 and in which AS UK are an equal partner. AS UK were Awarded Third Sector Excellence Award 2007 for the "Best use of the COMPACT" in ensuring the service remained user-led. Patients contact the charity mainly through the web site after receiving a diagnosis. AS UK family support workers visit the patient at home and make referrals to either the children's clinic if under 16 or the adult clinic. The Alström Service was hailed as a role model for rare disease by EURORDIS and other rare condition clinics followed our model. AS UK produced the first medical handbook on AS in 2004 dedicated to the son of the charity's founder and later also to her daughter. Both children did not survive heart transplantation. Today the charity supports 60 patients, their families, carers and the professionals who work with them. We produce information leaflets, flyers, quarterly newsletters and hold an Annual Family Conference. We have a dedicated Asian Support worker, supporting families where a culture of cousin marriages has led to increases in affected children. In 2010 ASUK were awarded a Big Lottery Medical and Scientific grant to develop a UK National Database and take skin cells from patients for further research. AS UK were presented with the EURORDIS Patient of the Year Award 2013 for outstanding services to Alström Patients. Please visit the following site for information and an aid to diagnosis: <http://youtu.be/mcrHbqHtktg>

Laurence-Moon-Bardet-Biedl society:

www.lmbbs.org.uk

The first official meeting of the Laurence-Moon-Bardet-Biedl Society (LMBBS) was held in 1989. Today, The Society supports over 400 families and communicates with over 150 health professionals involved in their care. The Society has produced many leaflets promoting the welfare of those with LMBBS, including a booklet aimed at the medical profession to provide a more in-depth look at the syndrome. The Society's latest publication 'Who Are We and How Can We Help You' is a leaflet about The Society itself and can be used to raise awareness of the syndrome and as a hand out for fundraising. There will soon be a new medical leaflet about genetic inheritance and cilia research. Our thanks go to all the clinicians for their assistance. The Annual Family Conference attracts many eminent and international speakers. The children and young adults benefit hugely from being part of a group where they are no different to anyone else. The newsletters and conference reports are intended to keep its members informed and supported, and hopefully ease the feelings of isolation that can occur with such a rare syndrome. Our helpline is also a lifeline to many, especially for the newly diagnosed, and is a chance to talk to someone who understands. In 1999, the society, together with Prof Beales, produced and sent out questionnaires to all its members. Prof Beales used the information from this study to produce the main diagnostic criteria, which are now used worldwide. In April 2010, after a successful bid with the National Commissioning service and input from Professor Phil Beales, the BBS Multi- Disciplinary Clinics commenced. These are held across four centres, and have brought about a major difference in how LMBBS is managed, with a focus on early intervention and good health management.

Wolfram syndrome UK:

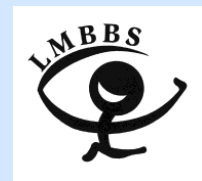
www.wolframsyndrome.co.uk

Paul and Tracy Lynch set up the support group after their daughter Jennifer was diagnosed with Wolfram syndrome in March 2010. The website is monitored by families affected by this rare genetic disorder and the aim is to raise as much awareness of the syndrome as possible. They feel that the more people that know about Wolfram syndrome, the better; and that as many doctors and health professionals as possible should be aware of this site. The charity also supports multidisciplinary clinics for children and adults across 2 hospitals. They have set up a forum to provide contact, support and an exchange of information between families in the UK who are affected by Wolfram syndrome; and an annual meeting for Wolfram syndrome



Living with Alstrom Syndrome:

<http://youtu.be/JV0n-ZcpOH0>



Self Management -
Laurence-Moon-Bardet
-Biedl Society :

http://www.youtube.com/watch?v=9f3_7zN2oVI



THE PATIENT EXPERIENCE

DANCING WITH BARDET-BIEDL SYNDROME

This story begins way back when I was 30 weeks pregnant with our oldest child Tom, where a routine scan showed that he had what they described as a kidney abnormality and I was told that this would have to be closely monitored after he was born. Tom arrived safely and was born with an extra finger on his left hand and an extra toe on his right foot. but none of the doctors or nurses seemed to be concerned about it. Tom's kidney function was monitored for the first few months of his life and we were told that it was fine. He was discharged from the kidney clinic aged 6 months, shortly after his extra digits had been removed.

Tom appeared a little lazy and his development was not as advanced as other children of his age. By the time Tom was 2 and a half he began attending pre-school and it was obvious that developmentally he was way behind his peers, although oblivious and in his own 'bubble of happiness'.

When Tom was 9 months old I was pregnant with our daughter Katie. Again I had a routine scan at 30 weeks and it showed the same kidney abnormality which Tom's scan had showed and sure enough she was born with an extra digit on her foot.

When Tom was 7 genetic testing revealed that Tom & Katie had BBS. Diagnosis day was life changing for me as a Mum, I really began to see the world through the eyes of the children and understand them and therefore be able to help them much more effectively, albeit on a rollercoaster journey.

Coping...

The two things which inspire me the most to carry on with strength and determination are Tom & Katie, our beautiful children. Two bright, funny, intelligent, kind and loving young people who are happy and have enriched our lives and made us proud parents. As far as coping is concerned, well there's not much option. To me it's not so much 'do we cope?' and more 'how do we cope?' by this I mean what tools do we use to make living with this thing as good as it can be. For me the answer is contained in one word – acceptance. I have learnt to totally accept Bardet-Biedl Syndrome. I have learnt to make room for it. I can't change it or make it go away, so I decided to accept it. People talk about fighting illness but to me this implies a winner and a loser. And we can never completely win with BBS and make it go away and losing is not an option, so to me it is like a dance. Sometimes I am not always a willing partner and during those quiet, reflective moments or those times when we are in a bad mood and tired, it would be easier to 'sit it out' . . . But if it wants to dance with me, then I must dance with it. I look at each appointment like a new routine, sometimes the steps are harder to learn than other times, but learn them I must if I am are to keep on top of this energetic thing called BBS.

Bardet – Biedl Syndrome never stops dancing... and nor must we

Emma Oates.

(This is an edited version of the full article which can be found at www.euro-wabb.org/en/presentations/euro-wabb-presentations)



THE PATIENT EXPERIENCE

Adorable twins Katie and Hannah were diagnosed with Alström Syndrome when they were 4 years old. Hannah was very ill for the first year of her life. After seeing a cardiologist at Leeds Hospital, the family were told she had a 33% chance of survival, a 33% chance of death and a 33% chance of needing a heart transplant. Both girls were diagnosed with nystagmus at 3 months old and parents noticed they both had an extreme sensitivity to light, after seeing many consultants the parents realised the severity of Katie and Hannah's sight loss. In 2010 parents Ian and Julie were referred to a geneticist as it was thought Katie and Hannah may have Alström Syndrome, this has now been confirmed. Julie Beck gives her thoughts:

"Since Katie and Hannah were diagnosed with Alström Syndrome, ASUK have provided both medical and emotional support. Without this support the last few years would have been even more difficult. Through ASUK's multi disciplinary clinics, we receive the specialist advice and treatments to enable the girls to lead as normal life as possible. As a family we look forward to the trips and annual conference, where we meet old friends and hope to make many new ones. ASUK gives us continued hope for the future."

The cheeky twins live life to the full and are always smiling and laughing together as well as keeping the Doctors on their toes!

Julie Beck



When I discovered the disease of my son (Wolfram syndrome) , I was alone in front of the screen of my computer. Three days before, life was so beautiful. He was diabetic since 4 years of age but he learnt to live with it and he was so happy. We visited an ophthalmologist and he discovered he had a bilateral optic atrophy.

I understood immediately that it was very serious. I undertook research on the internet and discovered Wolfram syndrome. A genetics consultant confirmed it few weeks later. I already wanted to create an association because I felt there was no research and no hope. I refused to stay and look the disease each day without doing anything.

Since I created The Association Syndrome de Wolfram, we have initiated research programs, The European registry, other studies on Wolfram syndrome

But another dimension is very important for the affected people: now they can share their experiences and advice, and they can discuss with other affected people. Some of them told me that they were waiting for this for several years. They felt so alone and not understood before.

Since the association was formed, they feel less alone and have a better life.

I want to continue my fight for all this patients...

Now I can sleep because I know I am doing all possible to help and save them.

Mdm Nolwen LE FLOCH



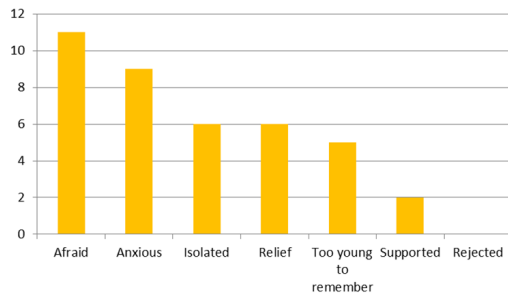
THE PATIENT EXPERIENCE



To inform the development of information resources, EURO-WABB researchers worked with Alström Syndrome UK (Kay Parkinson photographed left), Association Syndrome de Wolfram, the UK Wolfram syndrome association supported by Wellchild, and The Laurence-Moon-Bardet-Biedl Society to record patient experiences and identify the learning and information needs of affected

people and their families. The full report can be accessed via the project website.

Feelings Experienced at Diagnosis:



The most commonly reported feelings were those of fear and anxiety, with very few respondents feeling adequately supported. Most respondents felt the diagnosis had been given sympathetically, but a significant proportion wanted more explanation and counselling at the time of diagnosis.

Access to Information at Diagnosis:

'There wasn't much information, so I searched on the internet..'

About two thirds of respondents felt that they were not given enough information at diagnosis, with many seeking additional information on the internet. The reported experiences also differed widely with some patients feeling both informed and supported, and others less so. The main training requirements requested by families included: training in visual impairment; genetic counseling; healthy eating, exercise and lifestyle; mobility equipment; and dual sensory loss.

Recommendations for Health Professionals:

'Speak to each other and share information'

'Knowing more about the syndrome - we are the experts'

'Help with emotional / anxiety issues'

Four common areas for improvement have been identified by patients and families: the need to communicate better, both with fellow health professionals and with families; the need to be more knowledgeable about the conditions; to offer more support for psychological symptoms; and to provide more support for specific symptoms such as visual loss.

The EURO-WABB project is addressing these areas by working with family support groups to develop resources for comprehensive information, support and training; and with health professionals to improve coordination of care and communication between health professionals across subspecialties and hospital sites.

SCIENTIFIC ADVISORY COMMITTEE

The Scientific Advisory Committee is an expert multidisciplinary body that provides the rare disease community (clinicians, researchers, patient advocacy groups and industry) with independent and objective peer review of new research proposals that use data from the EURO-WABB registry.

Its goal is to support studies into the natural history of WABB diseases, genotype phenotype correlation studies, identification and validation of biomarkers, and future early phase intervention studies of new treatments.

Scientific Advisory Committee Members

- Ségolène Aymé, France
- Tim Barrett, UK
- Phil Beales, UK
- Andrew Hattersley, UK
- Chris Humphries, UK
- Nolwen Le Floch, France
- Miguel Lopez de Heredia, Spain
- Pietro Maffei, Italy
- Jan Marshall, USA
- Wojciech Mlynarski, Poland
- Virginia Nunes, Spain
- Richard Paisey, UK
- Véronique Paquis, France
- Kay Parkinson, UK
- Julia Rohayem, Germany
- Richard Sinnott, UK
- Vallo Tillmann, Estonia,
- Lisbeth Tranebjaerg, Denmark
- Fumi Urano, USA

Further details are available online at www.euro-wabb.org

ETHICS

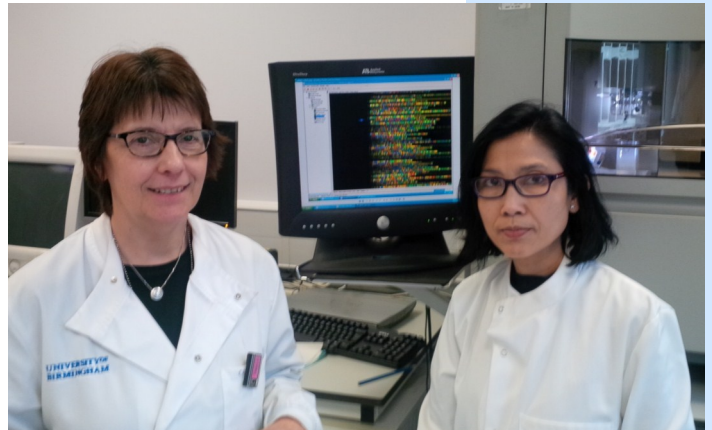
EURO-WABB has been designed to facilitate data sharing for maximum patient benefit. Participant consent includes additional optional data sharing with ethically approved national or international registries; agreement to be contacted about future research projects including clinical trials; and that any tissue samples collected or previously stored, may be used for in future ethically approved research projects.

Registry Consent Form

- I agree that my anonymised data can be included in a national disease registry for my condition **YES/NO**
- I agree that my anonymised data can be shared with other disease registries and research projects relevant to my condition within Europe **YES/NO**
- I agree that my anonymised data can be shared with international disease registries and research projects relevant to my condition that take place outside Europe **YES/NO**

GENETIC MUTATION DATABASE

Led by the University of Birmingham, Euro-WABB researchers and geneticists have established a series of mutation databases for the genes associated with WABB syndromes. These include Bardet-Biedl (18 genes), Alström (1 gene), Wolfram (2 genes), Wolcott-Rollison syndrome (1 gene) and Thiamine responsive megaloblastic anaemia syndrome (1 gene). Mutation data is linked to the citation report and published phenotypic data to allow basic genotype phenotype correlation.



Identified mutations have been compiled into open access databases using the Leiden Open access Variation Database (LOVD) software and are freely available online at: <https://lovd.euro-wabb.org>.

The databases currently contain information about 118 published *ALMS1* mutations, 430 mutations in BBS genes, 230 in Wolfram genes 80 mutations associated with other syndromes, together with the corresponding clinical phenotype where published. The databases are updated on a monthly basis.

Not only do the databases provide a useful resource for clinicians, it also ensures standardized reporting of genetic data into the project's patient registry increasing accuracy and cross-site analysis.

The mutation databases are registered and recognised by the HGVS and the project is a Human Variome Project Partner.

Dr Dewi Astuti and Dr Malgosia Zatyka, curators of the Euro-WABB genetic database at the University of Birmingham

LOVD Leiden Open Variation Database
EURO-WABB PROJECT OPEN VARIATION DATABASE
Wolfram syndrome 1 (wolframin) (WFS1)
Curators: Dewi Astuti and Professor Timothy Barrett

Home
Variants
Submitters
Submit
Documentation

WFS1 homepage Switch gene

LOVD Gene homepage

General information	
Gene name	Wolfram syndrome 1 (wolframin)
Gene symbol	WFS1
Chromosome Location	4p16.1
Database location	EURO-WABB Project, University of Glasgow
Curator	Dewi Astuti and Professor Timothy Barrett
Database reference for citations	[2]
PubMed references	View all (unique) PubMed references in the WFS1 database
Date of creation	May 27, 2011
Last update	March 18, 2013
Version	WFS1 130318
Add sequence variant	Submit a sequence variant
First time submitters	Register here
Reference sequence	Genomic reference sequence for describing sequence variants
GenBank reference	NM_006005.3
Exon/intron information	Exon/intron information table
Total number of unique DNA variants reported	227
Total number of individuals with variant(s)	390
Total number of variants reported	748
Subscribe to updates of this gene	

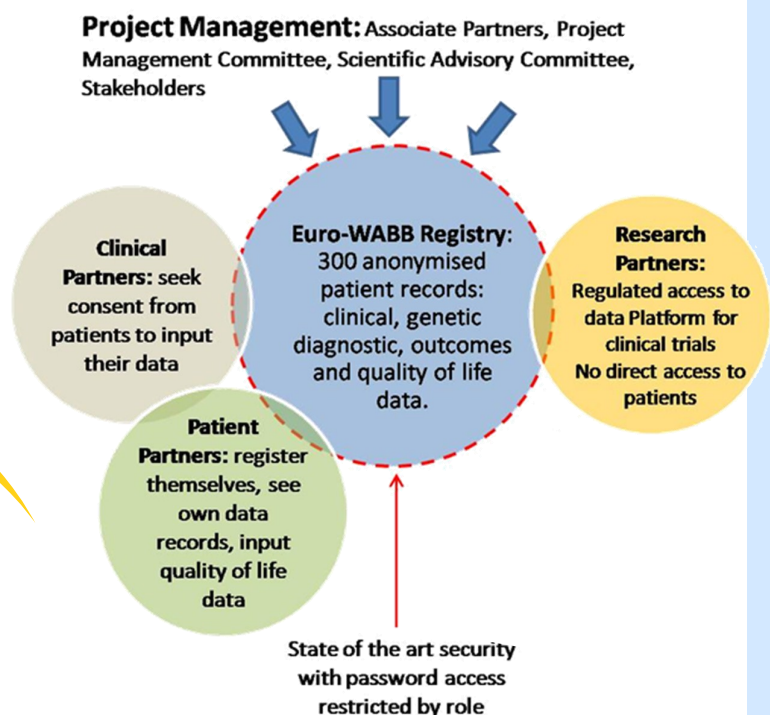
PATIENT REGISTRY

With nearly 300 participants recruited from more than 10 countries and across many recruitment sites, the Euro-WABB registry captures detailed clinical data about each syndrome. The registry includes data from the affected person and their clinician. All data is held in an anonymous format.

Registry data is split between a 'Core' (minimum), 'Extended' and 'Patient Experience' datasets. The core dataset includes 44 variables, consisting of summary current clinical and demographic data, and capturing the clinical and genetic data necessary to establish diagnosis. A further 370 variables capturing detailed phenotypic data are included in the extended dataset recording disease progression and date of onset of symptoms from the extended dataset. Patient experience records the patient's journey and their experiences from onset of symptoms, through diagnosis and the subsequent management of their condition.

The Registry allows longitudinal collection of data on individual participants, for instance from annual reviews. It also includes the facility for collection of imaging data.

Clinical diagnostic data across datasets is standardized using The International Classification of Diseases (ICD) coding system version 10. The endocrine subset data is further classified using The European Society for Paediatric Endocrinology (ESPE) Classification of Paediatric Endocrine Diseases. Data records are catalogued using a unique identifier, with only the clinician caring for the patient able to link the data record to the patient. The database offers secure access to, and sharing of, data at local, national and international levels.



STANDARDS OF CARE GUIDELINES

WABB syndromes, like many rare diseases, are often mis-diagnosed or subject to delayed diagnosis. Following diagnosis, clinical management requires the coordinated involvement of many different clinical specialties.

Together with clinical specialists for each syndrome, Euro-WABB has developed clinical management guidelines for health professionals for Wolfram, Alström and Bardet-Biedl syndromes, helping to ensure that patients receive a timely diagnosis and optimal clinical management and care.

These have been developed through multidisciplinary guideline development meetings, contribution and peer review by international experts in the fields, and advice from family support groups. Reassessment of existing and potential patients from The Registry have been used to refine the agreed diagnostic criteria and to facilitate the development of consensus referral, care and management pathways.

The development of these guidelines is identifying knowledge gaps and specific learning needs of healthcare professionals. In partnership with family support groups, educational materials and training tools are being developed to disseminate to target groups such as medical students and primary care professionals.

These guidelines are freely available via the website www.euro-wabb.org



“We got the results from Padova last week. In this way, I would like to thank you for your help with the testing and for the opportunity to join the EUROW-ABB project.”

Eszter Hegyi, MD
University Children’s Hospital,
Bratislava, Slovakia

GENETIC TESTING NETWORK

Equal access to genetic diagnostic testing

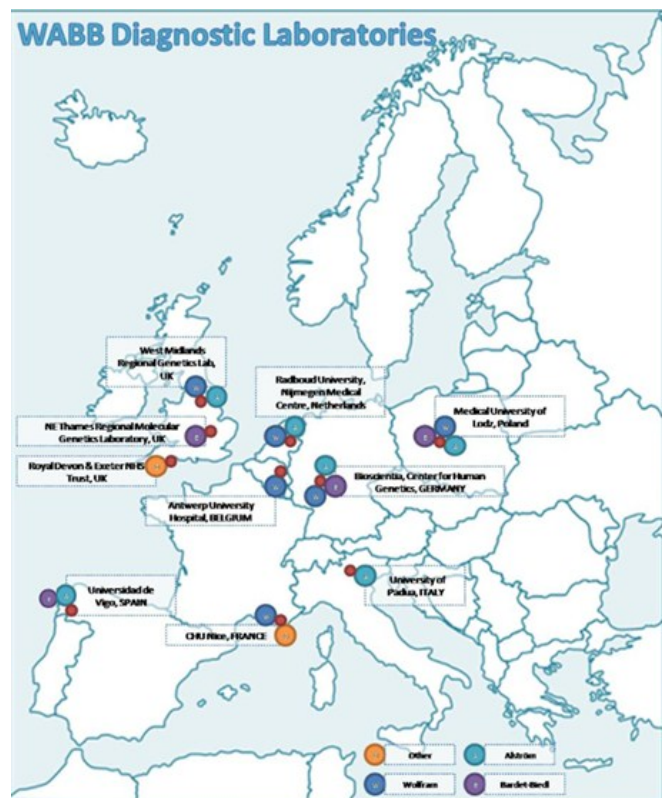
All three WABB syndromes are autosomal recessive conditions. Two causative genes have been identified for Wolfram, a single gene for Alström and multiple genes for Bardet-Biedl syndrome.

For registry participants who haven’t undergone genetic testing to confirm diagnosis, and where the cost of genetic testing is not met by national health funding, this is provided through the project. Access to diagnostic testing is via clinician referral, to ensure that any resulting diagnosis is coupled with appropriate counseling.

The Project has established a network of EU accredited and research laboratories that are able to offer genetic testing for WABB diseases. Information for each laboratory on the range of genetic tests offered, testing method, and reporting times is available on the EURO-WABB website.



In March 2012, Prof. Tim Barrett joined a team of specialists at the Huerca-Overa Hospital in Almeria, Spain for their multidisciplinary Wolfram syndrome clinic. Coordinated by Dr Gema Esteban-Bueno 16 individuals affected by Wolfram syndrome received specialist guidance for their condition. Dr Esteban Bueno also coordinates the ‘Asociación Nacional del Síndrome de Wolfram’ in Spain. (www.aswolfram.org). Genetic testing for these patients is now offered through the network.



Latest addition:
Wilhelm Johannsen Centre of Functional Genomics,
University of Copenhagen

CATALYZING NEW DEVELOPMENTS



Please select your preferred language

The EURO-WABB Project
An EU Rare Diseases Registry for Wolfram syndrome, Alström syndrome, Bardet-Biedl syndrome and other rare diabetes syndromes.
You are here: Home

EURO-WABB
The EURO-WABB Project is a collaboration of doctors, scientists and patient support groups from all over Europe. Within the EU Health Programme 2008-2013 and its call for promoting health through the creation of new registers for rare diseases, EURO-WABB is supported by The EU Directorate General for Health and Consumers (DG-SANCO) via its Executive Agency for Health and Consumers. The overall aim is for this register to be a key instrument to increase knowledge on these rare diseases, improve the lives of affected people through better management, and to develop clinical research.

Wolfram, Alström, Bardet-Biedl (WABB) and other Rare Diabetes Syndromes

[International Rare Disease Day is February 29th](#)

[News](#)

In bringing together a wide range of people with so many different disciplines, EURO-WABB is becoming a catalyst for new research proposals and spin-off projects, all of which can take advantage of the translational research platform provided by the Project. The following three examples of projects using the EURO-WABB resources contribute to international efforts to identify new treatments for these diseases:

Wolfram syndrome

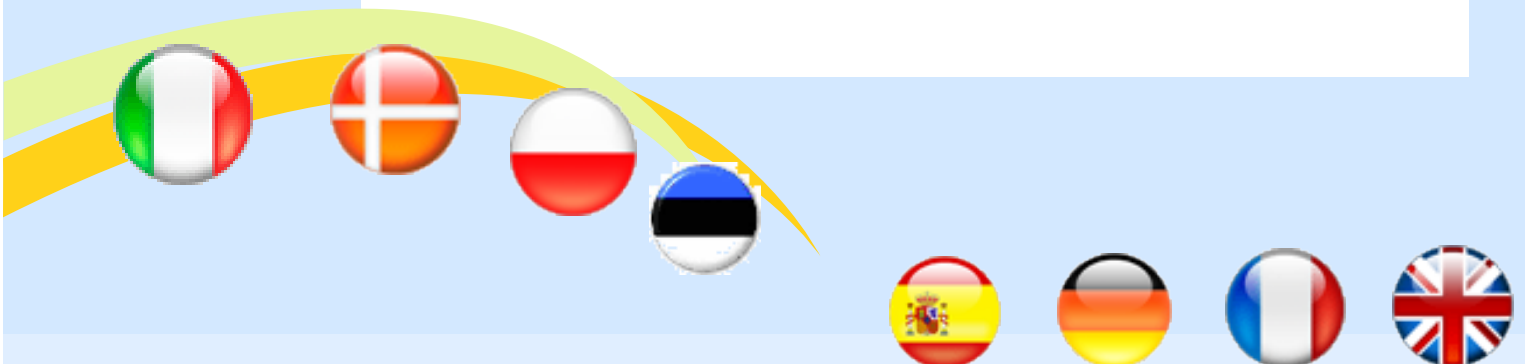
Association Syndrome de Wolfram is supporting a drug repurposing project to identify licensed medicines which may slow down or halt the neurodegeneration in Wolfram syndrome. They also initiated a research program on gene therapies for the eye, led by a French team and bringing together international collaborators. At the same time Professor Fumi Urano is establishing a consortium to commence clinical trials; and both groups have identified similar classes of drugs. The EURO-WABB Registry will be the portal of entry for recruitment of European patients to an international clinical trial of new treatments for Wolfram syndrome.

Alström syndrome

A European Union Innovative Medicines Initiative (IMI) grant has been awarded to a consortium of European researchers and pharmaceutical companies to develop stem cells developing novel drugs for common diseases (STEM cells for Biological Assays of Novel drugs and predictive toxicology). This includes a work package developing stem cell models of diabetes. There is an opportunity within this work package to develop stem cell models for Alström syndrome as a monogenic model of insulin resistant diabetes (and Wolfram syndrome as insulin dependent diabetes). The ethics approval for EURO-WABB is allowing early collection of skin biopsies from participants into biorepositories for de-differentiation to iPSCs.

Bardet-Biedl syndrome

Professor Beales (UCL, London, UK) is leading current research efforts with an industrial partner to develop clinical trials of gene editing technology. This is of particular relevance to about one third of Bardet-Biedl patients who carry premature stop mutations in their genes. There is also ongoing interest in new treatments for the kidney disease. The development of the EURO-WABB registry of over 100 consenting participants offers a unique resource from which to invite participants to clinical trials.



EURO-WABB—THE FUTURE

The platform EURO-WABB has established for establishing the natural history of these diseases, developing international cohorts, consensus management guidelines, open access genetic databases, and improved access to genetic testing, is accelerating translational research. The Project is providing a platform to support new developments across the field, and is proving its value to the rare diseases community. Its coordinator, associate and collaborating partners are committed to sustaining this project in the long term.

The future will see the Project further develop this essential infrastructure, playing an increasing role in areas such as long term assessment of clinical care pathways, validation of disease monitoring tools, lobbying activities to advocate on behalf of affected people, and updating and implementing of care standards across the spectrum of these diseases. We will support benchmarking of service delivery against care standards, supporting idea generation from leaders in the field, and supporting training and education to develop new care and trial sites with the expertise to treat these rare diseases.

We hope that our efforts will ensure that the rare disease community can move forward together to develop the treatments for which affected people, their families and their health professionals have waited for so long.

A number of local investigators have recruited participants for the registry. We are immensely grateful to:

- Philip Beales, UK
- Gema Esteban Bueno, France
- Birute Burnyte, Lithuania
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- Kaire Heilman, Estonia
- Olga Liaugaudiene, Lithuania
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- Julia Rohayem, Germany
- Vallo Tillmann, Estonia
- Donald Whitelaw, UK
- Agnieszka Zmyslowska, Poland



ACKNOWLEDGEMENTS

We would like to thank the patient support groups for their kind help in compiling this brochure; and the families that it is our privilege to know.

In addition to funded partners, a number of other individuals and organisations have also been instrumental in the design, development and success of the project.

COLLABORATING PARTNERS

Bispebjerg Hospital, DENMARK (Lisbeth Tranejaerg)

University of Münster, GERMANY (Julia Rohayem)

Association Syndrome de Wolfram, FRANCE (Virginie Picard & Nolwen LE FLOCH)

CIBERER, SPAIN (Miguel Lopez de Heredia)

University of Central London, UK (Philip Beales)

LMBBS, UK (Chris Humphries)

South Devon Healthcare NHS FT, UK (Richard Paisey)

ISPAD Rare Diabetes Registry, International (Andrew Hattersley)

University of Washington, USA (Fumi Urano)

The Jackson Laboratory, USA (Jan Marshall)

Spanish association for the research and help of Wolfram's syndrome, SPAIN (Gema Esteban Bueno & Luisa Maria Bottelli)

University of Nice, FRANCE (Annabelle Chaussenot)



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