



EURO-WABB

An instrument to increase knowledge, improve the lives of affected people through better management, and to develop clinical research

Kay Parkinson
CEO Alström Syndrome UK
EURO-WABB Associate Partner

Wednesday 31st October 2012
Council of European Federations 5th Workshop



From Parent



To Partner



Kay Parkinson, Founder & CEO Alström Syndrome UK
Associate Partner, EURO-WABB



Alström Syndrome UK
Support Group



Matthew
Born 9/02/1978 -
Diagnosed with Alström
Syndrome in 1996- age 18

Charlotte
Born 11/04/1981-
Diagnosed with Alström
Syndrome in 1996-age 15

Symptoms

- Childhood blindness-intense photophobia and nystagmus - very visible signs –
- Lead to five different diagnosis of their eye condition being made.
- Dilated cardiomyopathy-collapse at 11 weeks in heart failure-no connection?
- Hearing loss- misdiagnosed as a learning difficulty
- Obesity- mums fault for over feeding!
- All these conditions are deemed to be un-connected and treated separately
- For 18 years we are isolated, alone and undiagnosed
- Labelled “Disabled”- Diagnosis came by chance -

Actions & Achievements

- Alström Syndrome UK Charity started in 1998 (www.alstrom.org.uk)
- Unification of affected UK families through our strong support group
- Development of patient led NHS funded multi disciplinary clinics
- A role model for clinics for other rare diseases such as **Bardet-Biedl** and **Wolfram** syndrome
- Patient led- research funded by the Big Lottery Charities Board
- Medical handbook, patient information leaflet and newsletter produced
- Euro-WABB Rare Diseases Registry..



EURO-WABB: An EU Rare Diseases Registry to support efficient diagnosis, treatment, and research for Wolfram, Alström, Bardet-Biedl and other Rare Diabetes Syndromes in Europe

In 2011, Alström Syndrome UK (ASUK) invited to join EURO-WABB (EURORDIS recommendation) to lead the project's dissemination work package.

- ASUK is the only patient group partner
- ASUK join an EU Registry of patients - increased visibility
- Opportunity to find new contacts and connect with more families and share our work
- Collaboration with allied conditions increases interest in all WABB diseases
- Sharing our knowledge and experience benefits more families

The WABB diseases



Wolfram syndrome

Childhood diabetes, optic atrophy, deafness, neuro-degeneration



Alström syndrome

Obesity, retinal dystrophy, deafness, often cardiomyopathy and diabetes

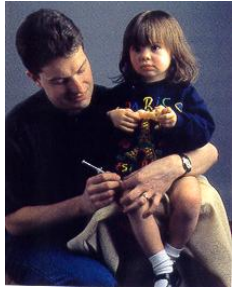
- Wolcott-Rallison syndrome
- Roger's syndrome
- Other even rarer syndromes



Bardet-Biedl syndrome

Obesity, retinal dystrophy, deafness, often diabetes, polydactyly, cystic kidneys

The Problem: Different combinations of symptoms



Diabetes

Common diabetes

Isolated vision impairment

Vision Loss



Non-syndromic deafness



Hearing loss

Cardiomyopathy

Isolated cardiomyopathy



Obesity

Common obesity

WARNING !!!

- ⚠ Failure to diagnose patterns
- ⚠ Too rare for doctors to learn
- ⚠ Little or no research
- ⚠ Inadequate patient information
- ⚠ Failure to treat properly

WARNING!!!



Project Overview



- EU Funded 3 year project (Health Programme Framework)
- 9 Associate Partners across Europe
- European Rare Diseases Registry collecting clinical and diagnostic data
- Wolfram, Alström and Bardet-Biedl (WABB) syndromes and other syndromes such as Wolcott-Rallison and Thiamine-Responsive Megaloblastic Anaemia (TRMA)
- Project activities divided into 6 work packages



EURO-WABB Project Aims

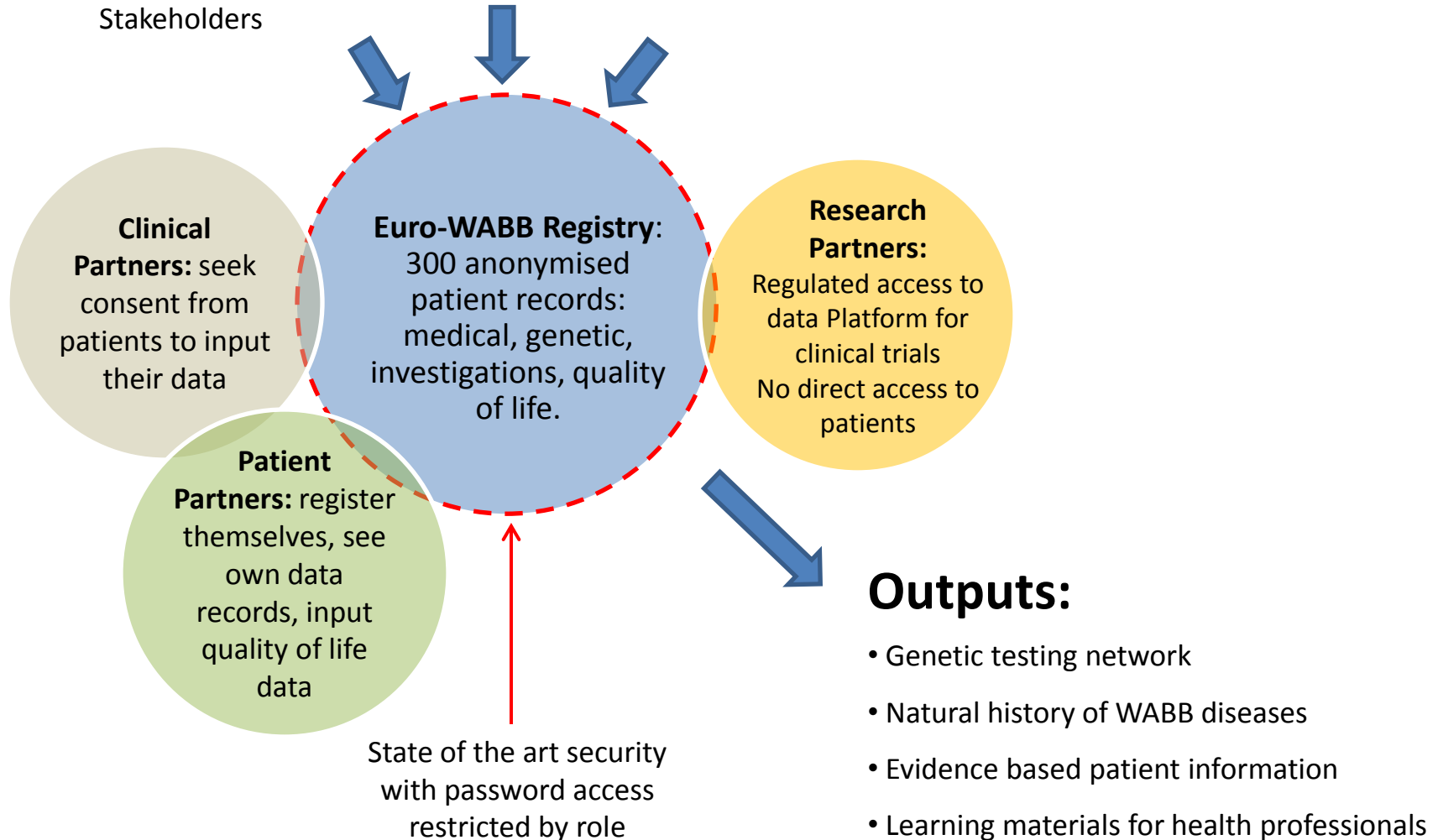


- EU registry of clinical, molecular genetic and outcome data on up to 300 affected patients for each condition
 - To establish disease characteristics, best management, and outcomes
 - To assess effectiveness of clinical management and quality of care
 - To provide a cohort of interested patients for future clinical studies
 - To understand relations between clinical and genetic findings
- High usage by:
 - Access to rapid, free genetic testing
 - Up to date accurate patient information
 - Teaching resources for health professionals

Euro-WABB Registry



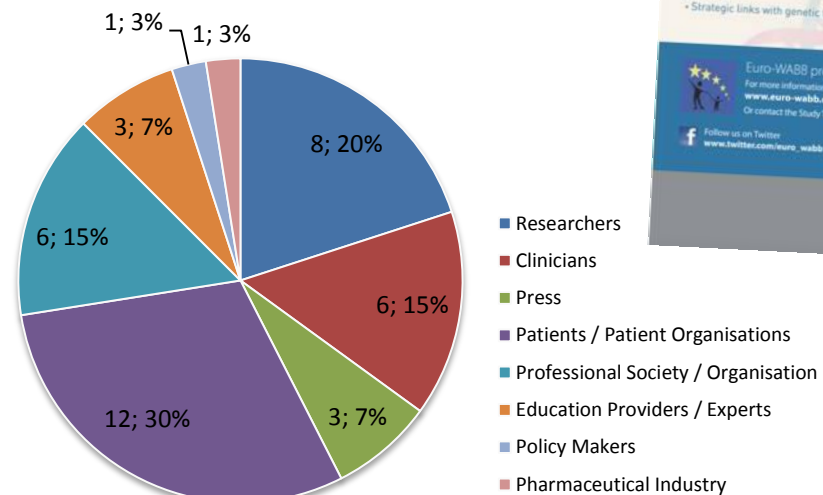
Project Management: Associate Partners, Project Management Committee, Scientific Advisory Committee, Stakeholders



Work Package 2: Dissemination



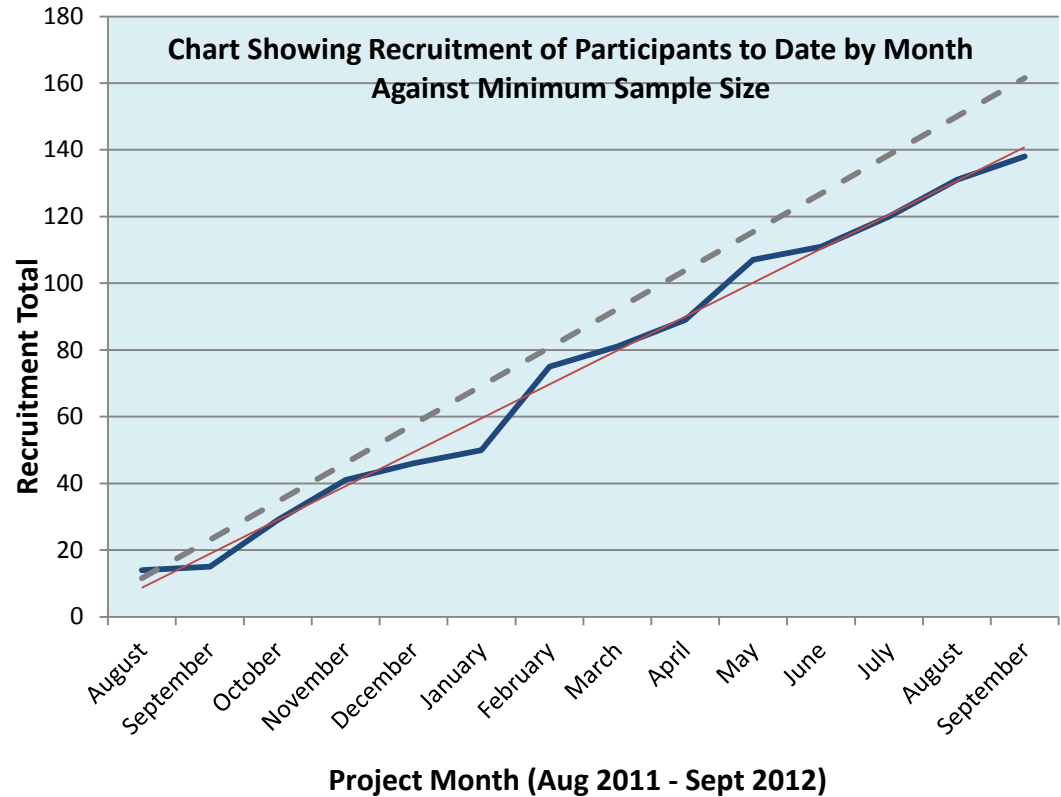
- Identifying and engaging with stakeholders
- Awareness Raising
- Associate Partner & Member of the Project Management Committee contributing to the delivery of Euro-WABB
- Leading analysis of the learning and information needs of patients and families
- Contributing to the development of guidelines



Recruitment

- Ethics Approvals in 8 Member States
- 138 Participants recruited
- Identification of new clinical partners ongoing

Member State	Ethics Approval
Poland	14/06/2011
UK	20/06/2011
Italy	14/07/2011
Estonia	17/10/2011
Spain 1	24/11/2011
France	18/04/2012
Slovakia	16/05/2012
Spain 2	30/05/2012
Germany	03/08/2012
Denmark	<i>Pending</i>
Greece	<i>Pending</i>
Lithuania	<i>Pending</i>



	France	Italy	Poland	Slovakia	UK	Spain
Alström	0	15	4	1	10	0
Wolfram	19	4	20	0	19	1
Bardet-Biedl	0	0	0	0	37	0
Wolcott-Rallison	0	0	0	0	1	0
TRMA	0	0	0	0	0	0
Total	19	19	24	1	74	1
Grand Total	138					

Sustainability



- **Drivers:**
 - Utility to health professionals, researchers and patients
- **Plans for continuation:**
 - EU health providers: network of experts in WABB diseases
 - Industry: new indications for existing drugs
 - Disease specific charities
- **Future developments:**
 - Alström Europe and later possibly European Wolfram and BBS Federations
 - I-WABB – International registry for WABB diseases
 - Patients from Morocco, Saudi Arabia, Iran

Research projects using the EURO-WABB resource



- **Scientific Advisory Committee**
 - Comment on applications to access data
 - Assess scientific and potential for benefit to patient group
- **Pharma and Academic Researchers**
 - Treatment studies
 - Raise awareness and encourage applications from researchers
- **Research Ideas**

Acknowledgements:

Mathew Parkinson 9/02/78 to 11/04/2003

Charlotte Parkinson 11/04/81 to 29/04/2010

John Parkinson (Husband) supportive throughout
The families and patients of Alström Syndrome UK

EURORDIS

Associate Partners

www.euro-wabb.org



Inserm



www.euro-wabb.org

Thank you



Alström Syndrome UK
Support Group



UNIVERSITY OF
BIRMINGHAM



IDIBELL
Institut d'Investigació Biomèdica de Bellvitge