NAME OF PROJECT:
EURO-Rare Diabetes: an EU rare diseases registry for rare diabetes syndromes including Wolfram, Alstrom and Bardet Biedl syndromes (EURO-Rare Diabetes).

CONTRACT NUMBER:
2010 12 05

WEBSITE ADDRESS:
To be confirmed

PARTNER COUNTRIES:
Main partner: United Kingdom
Associate partners (funded by The Action): Estonia, France, Italy, Poland, Spain.
Collaborating partners (not funded by The Action): Denmark, Germany, Japan, Romania, USA.

LENGTH OF FUNDING:
36 months

FUNDING AGENCY:
DG Sanco

SUMMARY:
Wolfram, Alstrom and Bardet Biedl (BBS) (WABB) syndromes (0.57, <0.2 and 0.8 cases/100,000 respectively; Orphanet Report Series Nov 2009 No. 1) are rare genetic diseases with clinical overlap (they can all cause diabetes mellitus or intolerance of the body to glucose), chronically debilitating, and highly complex. Affected people are distributed throughout the EU but disproportionately affect ethnic minorities. The diseases may progress to death in early adulthood. There are as yet no orphan drug treatments available, and no access to well characterized cohorts of patients. The diseases show insights into common conditions (e.g. Wolfram variants and type 2 diabetes; Alstrom/BBS and obesity), but research is scarce and scattered in different laboratories throughout the EU. The lack of specific health policies for these diseases and the scarcity of expertise, translate into delayed diagnosis and difficult access to care. There are almost no multidisciplinary teams of experts in these diseases. Genetic testing centres are concentrated in a few member states, and there is unequal patient access to testing across the EU.

The idea for this project was from Nolwenn Jaffre, President of the French Wolfram Association, who brought together Wolfram syndrome researchers in Paris in October 2009. The final application brought together researchers in Alstrom syndrome, Bardet Biedl syndrome, and Alstrom Syndrome UK as partners. The general objective of this project is to support efficient diagnosis, treatment, and research for the overlapping rare genetic diseases Wolfram, Alstrom and Bardet Biedl syndromes and other rarer diabetes syndromes in Europe. We will achieve this by implementing an EU registry for Rare Diabetes Syndromes (RDS), containing anonymised clinical, genetic diagnostic and outcome data. The purpose of the registry is: a) to establish the natural history of RDS (their characteristics, management and outcomes); b) to assess clinical effectiveness of management and quality of care; c) to provide a cohort of families interested in research who can be approached through their local doctors for participation in treatment studies; d) to establish if the genetic mutations can predict the complications or response to treatments. We will aim for rapid, readily available genetic testing; and up to date, accurate patient information, FAQS, and education material for health professionals.

We will ask local doctors to approach affected families for consent to include their anonymised clinical data record in The Registry. The local doctor will control access to the records that he/she adds to the Registry. Researchers will then apply to an oversight committee for permission to view the data records. If there is a new clinical trial of a treatment, the researcher will have to go through the local doctor for permission to approach his or her patients.

We hope this will lead to an increase in the volume and quality of clinical research in RDS diseases. RDS diseases will have increased visibility to the research and health provider communities through Orphanet and EURORDIS. We will also use the anonymised data records to develop international guidelines on management of the diseases, and use the registry to assess the clinical effectiveness and cost-effectiveness of standard care and new interventions in a real-world setting. This will hopefully lead to improvements in quality of care. The Registry will also identify disparities between health care outcomes and provide evidence for health service providers for improvements.

If you are reading this and either look after someone with one of these syndromes, or you or a member of your family are affected, then please do get in touch, either directly to me by email (t.g.barrett@bham.ac.uk) or via our website (www.euro-rare-diabetes.....). We will add you to our mailing list and send you regular news updates on how the project is progressing and how to get involved. Thanks for reading this!