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An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome

JA2015 - GPSD [705038]

START DATE: 01/01/2011

END DATE: 01/07/2014

DURATION: 42 month(s)

CURRENT STATUS: Finalised

PROGRAMME TITLE: Second Programme of Community action in the Field of Health 2008-2013

PROGRAMME PRIORITY: -

CALL: Promote Health (Hp-2010)

TOPIC: PROMOTE HEALTH (HP-2010)

EC CONTRIBUTION: 900000 EUR

KEYWORDS: Diagnostic, Health system, Patients registries, Quality evaluation, Rare diseases and disorders

PORTFOLIO: Rare diseases

General objectives

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 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 an inventory of patients for recruitment to intervention studies; d) to establish genotype-phenotype correlations. We will achieve high usage of the registry by linking it to rapid genetic testing; and to up to date, accurate information, FAQS, and education material.

Strategic relevance and contribution to the public health programme

This supports equal access to genetic testing, education of health professionals, and empowerment of patients. (Council Recommendation on rare diseases); adequate inventorying of RDS diseases (Section II); supporting research (Section III); development of centres of expertise (Section IV); gathering expertise at European level (section V); empowering patient organizations as partners (Section VI); developing sustainability by underpinning a future European Reference Network for RDS diseases (Section VII); supporting the High Level Pharmaceutical Forum Recommendations (2008); and supporting improvement in health outcomes, a key Lisbon Strategy indicator. The contribution to the programme is through: a) increased knowledge on these rare diseases by pooling together data on larger number of patients; b) support for research by allowing access to investigators for epidemiological, clinical, genetic and interventional studies; c) effective dissemination of results via Orphanet; d) advocacy for improved quality of services via EURORDIS; e)balanced participation.

Methods and means

Methods and means. We will use validated, quantitative questionnaires and focus groups of health professionals, to scope the support requirements of centres for submitting data to the Registry. We will develop a consensus on a core dataset for the Registry, then develop a multifunctional web based Registry with user friendly browser-based access. We will create a RDS microarray capable of identifying up to 600 different mutations. We will undertake quantitative questionnaires and focus groups for patients and health professionals to compile their learning and information needs; write education material and patient information on RDS diseases, and use it to support `meet the expert` platforms, and fora for client groups.

Expected outcomes period

Expected outcomes. There will be a step change in volume and quality of clinical research in RDS diseases. The registry will be also be transferable to scientists exploring the mechanisms underlying common diabetes and obesity. This will change our understanding of these rare diseases through increased knowledge of the natural history and genotype phenotype relations informing prognosis. RDS diseases will have increased visibility to the research and health provider communities through Orphanet and EURORDIS. There will be a change in clinical effectiveness of services for RDS patients. The registry will provide data for assessing the clinical effectiveness and cost-effectiveness of standard care and new interventions in a real-world setting. This will lead to improvements in quality of care. The Registry will identify disparities between health care outcomes and provide evidence for health service providers for improvements.

COORDINATOR, LEADER CONTACT AND PARTNERS

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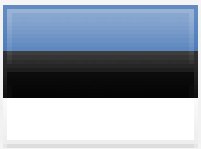

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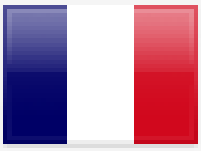

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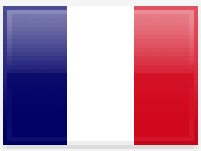

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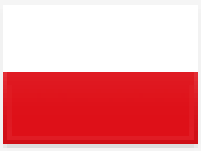

Medical University of Lodz
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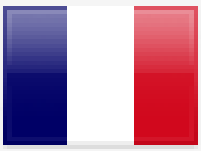

National Institue of Health and Medical Research (INSERM)
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Country: France

OUTPUTS

D01 - 01 - Final report (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/euro-wabb_final_technical_report_-_final_version.pdf

D01 - 02 - Final report Appendix (EN)

University of Birmingham C/O Diabetes Unit
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Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/appendix_.pdf

D01 - 03 - Leaflet (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d01_01_lft_en_ps.pdf

D01 -01 - Interim report (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d01_02_ir1_en_ps.pdf

D01 -02 - Interim report 2 (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)

Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d01_03_ir2_en_ps.pdf

D01 -03 - Report on achivement (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d01_04_oth_en_ps.pdf

D01 -04 - Leaflet (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d01_05_oth_en_ps.pdf

D02 - 01 - Scoping evaluation report of project (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d02_01_oth_evr_ps.pdf

D02 - 02 - External evaluation plan (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d02_02_oth_en_ps.pdf

D02 - 03 - Activities_CV (EN)

University of Birmingham C/O Diabetes Unit
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http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d02_03_oth_en_ps.pdf

D02 - 04 - Letter (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d02_04_oth_en_ps.pdf

D03 - Scoping Exercice & reports (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d03_00_oth_en_ps.pdf

D04 - 01 - Development of a web based registry (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d04_01_oth_en_ps.pdf

D04 - 02 - Development of a web based registry report (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)

Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d04_02_oth_en_ps.pdf

D05 - Publication of consensus management guidelines and datasets (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d05_all_oth_en_ps.pdf

D06 - Input of datasets on at least 300 patients with RDS disease (100 for each main disease) (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d06_00_oth_en_ps.pdf

D07 - Development of an EU Genechips for genetic testing (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d07_00_oth_en_ps.pdf

D08 - A report on the learning needs of health professionals (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and

Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d08_00_oth_en_ps.pdf

D09 - Development of health professional education and patient information material to support the Registry (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d09_01_02_oth_en_ps.pdf

D10 - Delivery of an EU workshop to disseminate the findings at the end of the project (EN)

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_d10_00_oth_en_ps.pdf

Final Report

University of Birmingham C/O Diabetes Unit
An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (EURO-WABB)
Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/euro-wabb_final_technical_report_-_final_version.pdf

Internal Assessment

University of Birmingham C/O Diabetes Unit
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Published on: 01/01/2011
http://azorina.cc.cec.eu.int:8082/publications/20101205/20101205_euro_wabb_assessment-note.pdf