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European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability

JA2015 - GPSD [705038]

START DATE: 01/03/2018

END DATE: 28/02/2019

DURATION: 12 month(s)

CURRENT STATUS: Finalised

PROGRAMME TITLE: 3rd Health Programme (2014-2020)

PROGRAMME PRIORITY: -

CALL: EUROPEAN REFERENCE NETWORKS SPECIFIC GRANT AGREEMENTS COVERING YEAR 2018

TOPIC: ERN Specific Grant Agreements Year 2

EC CONTRIBUTION: 140998 EUR

KEYWORDS: Birth Defects, Congenital Malformation, Disability, Intellectual, Intellectual Disability, Multiple Anomaly Syndrome, Rare Chromosome Disorder

Project abstract

Birth defects may result in death, chronic illness or long term disability with a significant impact on individuals, families,
health care systems and societies. Many birth defects are also associated with intellectual disability as part of rarer patterns
or syndromes and require multidisciplinary care. More than 8000 rare syndromes have been described. Thus although rare
these are collectively important conditions. They may have genetic, environmental and multifactorial causes but in 50% of
cases the cause is currently unknown. Identifying causes and studying the natural history of multiple anomaly syndromes
guides management or treatment, provides answers for families and healthcare professionals, furthers understanding of
normal development through research and may lead to prevention. All of these will have significant health economic benefits
and guide commissioning of future services. However, expertise in the study of these conditions is limited to a few experts,
in major healthcare centres. In some EU member states the specialty of syndrome diagnosis and management is not well
established, there is no specialist training and there are few clinical and laboratory resources.

We propose to continue the patient-centred European Reference Network for Rare Malformations and Intellectual Disability, ITHACA, which aims to meet the needs of patients, both diagnosed and undiagnosed. ITHACA works to improve access to diagnostic expertise by utilizing an innovative telehealth approach and guiding quality assured diagnostic testing.

Work package

Work Package 1: Management

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Link together centres for RCMCID for the benefit of patients
* Have a robust process in place to monitor activity
* Interact with other networks to improve quality/safety of care
* Create a forward-looking network with future potential

Work Package 2: Dissemination

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Communicate effectively making use of IT
* Improve provision of resources to patients
* Share experience of the added value of ERNs

Work Package 3: Evaluation

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Build an ERN with high quality services
* Improve equity of access for patients and trainees in less well developed MS
* Add value to patient care

Work Package 4: Expert Patient Care (formerly Guidelines Management)

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Agree rare disorders and malformations where guideline required
* Evaluation of existing guidelines against AGREE II
* Improve patient care through use of high quality guidelines
* Put in place a process for new European guideline development

Work Package 5: TeleHealth Diagnosis

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Share diagnostic expertise for direct patient care
* Develop TeleHealth approaches for patient care
* Use Telehealth to identify and study new rare diseases

Work Package 6: Patient Registries

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Identify and utilise registries for RCMCID for the benefit of patients
* Interact positively with registries to improve data quality
* Promote connections registries/biobanks
* Promote networking of disease-specific national patient registries
* Demonstrate improved patient care through use of EU registries

Work Package 7: Research

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Through establishment of the ERN, enhance research
* Actively involve patients in the research agenda of the ERN
* Share research knowledge and expertise through teaching and training
* Generate research findings from our collaborative working

Work Package 8: Teaching and Training

Start month: 1
End month: 12
Work Package Leader: CMFT NHS TRUST

* Define currently available education and training resources for professionals/families
* Identify patients’ and professionals future education/training needs
* Rapid dissemination of NEW information
* Ensuring training needs for the future are met
* Improve access to educational resources

COORDINATOR, LEADER CONTACT AND PARTNERS

COORDINATOR

MANCHESTER UNIVERSITY NHS FOUNDATION TRUST (CMFT NHS TRUST)

Nowgen Centre, Grafton Street, M13 9WU
Manchester

United Kingdom
WEBSITE: http://www.mft.nhs.uk

PARTNERS

No partners related to the current project

OUTPUTS

Implement Network Evaluation Plan

CMFT NHS TRUST
European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability (ERN-ITHACA)
Published on: 31/07/2019
Evaluation plan written by Management team and agreed at Board level EP reviewed throughout year, with summary/analysis included in Year 2 Technical Report

Agreed high quality strategy, including data set, for patient registry agreed

CMFT NHS TRUST
European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability (ERN-ITHACA)
Published on: 31/07/2019
Registries WP to lead scoping measures exercise, then propose, agree and circulatestrategy.

Results of patient survey on research

CMFT NHS TRUST
European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability (ERN-ITHACA)
Published on: 31/07/2019
Research WP to draft survey and circulate to Patient Groups. Results collated, discussed and circulated/uploaded to core IT platforms

Report from patient research group

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Published on: 31/07/2019
Interested parties contacted, first call/meeting held

Completed education/training needs survey

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European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability (ERN-ITHACA)
Published on: 31/07/2019
Results from Teaching and Training survey discussed and circulated/uploaded

e-Learning module developed

CMFT NHS TRUST
European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability (ERN-ITHACA)
Published on: 31/07/2019
Software licence purchased, staff trained, module developed by PM team and Coordinator.

List of meeting reports from high quality meetings and conferences which have been published on the website

CMFT NHS TRUST
European Reference Network For Rare Congenital Malformations and Rare Intellectual Disability (ERN-ITHACA)
Published on: 31/07/2019
WP leaders compile list, circulate and ITHACA members write reports of meetings, send to PM team, reports uploaded throughout the year