**EURO-WABB PROJECT:**
EURO-WABB is a 3 year project with funding from the EU Health Programme Framework. The general objective of the project is to support efficient diagnosis, treatment and research for Wolfram, Alström and Bardet-Biedl (WABB) syndromes and other rarer diabetes syndromes in Europe (Wolcott-Rallison syndrome, and Thiamine-responsive megaloblastic anaemia) through the development of a EURO-WABB European Registry of patients.

**RARE DIABETES SYNDROMES:**
WABB syndromes are all rare autosomal recessive disorders. They are chronically debilitating, highly complex, and often subject to misdiagnosis and non-diagnosis. Access to molecular genetic testing is unequal between European citizens and treatable complications are often missed. There are no orphan drug treatments available, and no access to well characterized cohorts of patients to undertake research. The syndromes exhibit clinical overlap; all can cause blindness, deafness, and diabetes mellitus or impaired glucose tolerance. Wolfram syndrome is characterized by young onset diabetes and bilateral optic atrophy. It is also known as DIDMOAD syndrome, for the other disease features including diabetes insipidus, diabetes mellitus, optic atrophy, and deafness. Alström syndrome is characterised by infancy onset obesity and retinal dystrophy. The condition is first apparent as photosphobia, nystagmus, leading to progressive visual loss from childhood. Obesity, insulin resistance, and partial nerve deafness follow, then type 2 diabetes (80%), severely high blood triglycerides, cardiomyopathy (50%), with more than 25% affected in infancy and renal failure. Bardet-Biedl syndrome is characterised by polydactyly, infancy onset obesity and retinal dystrophy. Associated features include early blindness, cystic kidneys, renal failure, global learning difficulties, urological problems, and neurological deficits including sensorineural deafness.

**REGISTRY DATASETS:**
We agreed a common dataset of clinical, investigation and molecular diagnostic data to distinguish between the syndromes and provide a searchable database for researchers. We also incorporated data fields for Wolcott-Rallison, Thiamine-responsive megaloblastic anaemia (TRMA) deafness and diabetes, and other rarer diabetes syndromes. We wrote an ethics submission template for national approvals, which includes consent to link national and international registries. The core dataset includes 44 data fields which define and separate the syndromes; the extended dataset comprises 370 fields of detailed phenotyping information. The core dataset has been designed to be completed in 15 minutes. The registry data is standardised using ESPE and WHO ICD-10 coding systems where possible. The full datasets can be accessed at [www.euro-wabb.org](http://www.euro-wabb.org).

**DATA COLLECTION:**
We designed a web-based registry (NeSC, University of Glasgow) with built in security for data confidentiality, anonymised data collection, and facility for patients to self register. We undertook a scoping exercise to identify barriers to data entry and this has informed the development of the registry interface and the support available to those entering data.

**RECRUITMENT:**
We currently have ethics approval in 6 EU member states, and the first 82 participants consented, from Italy, Macedonia, Poland, Turkey, and UK. Ethical approvals are underway in Germany and France. The recruitment target is 300 participants. If you are interested in taking part, please email: euro-wabb@bch.nhs.uk

**DISCUSSION:**
This EU Registry will improve patient services, health professional awareness, and allow recruitment into clinical trials. The common core dataset for European Union states can be shared between national rare disease registries as they are developed; and will allow linkage with other international disease registries. The registry data will be used to increase the understanding of the natural history of WABB diseases, to serve as an evidence base for clinical management, and to aid the identification of opportunities for intervention to stop or delay the progress of the disease processes. The detailed clinical characterisation will allow inclusion of patients into studies of novel treatment interventions, including targeted interventions in small scale open label studies; and enrolment into multi-national clinical trials. The registry will also support wider access to genetic testing, provide information for affected families and health professionals; and encourage international collaborations for patient benefit. It is hoped that this will lead to an improved quality of life for WABB patients achieved through earlier diagnosis, prompt identification and management of complications and increased healthy life years.

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**EURO-WABB ASSOCIATE PARTNERS**
(1) Birmingham Children’s Hospital, UK; (2) National Institute of Health and Medical Research (INSERM), France; (3) Università degli Studi di Padova (UNIPD), Italy; (4) University of Glasgow (NeSC), UK; (5) Medical University of Lodz (MUL), Poland; (6) Fundació Institut Investigació Biomèdica de Bellvitge (IDIBELL), Spain; (7) Centre Nationale de la Recherche Scientifique (CNRS), France; (8) Alström Syndrome UK (ASUK), UK; (9) Universitätsklinikum Münster, Germany; (10) University of Tartu (TU), Estonia; (11) Bispebjerg Hospital & University of Copenhagen, Denmark; (12) University of Birmingham (UB), UK.