ALL A11

Ministero della Salute Direzione generale della ricerca e dell'innovazione in sanità **PNRR: M6/C2_CALL 2022 Letter of intent**	Finanziato dall'Unione europea NextGenerationEU
Project Code: PNRR-MR1-2022-12376596	**Call section:** Malattie Rare
Applicant Institution: Veneto	**Applicant/PI Coordinator:** Burlina Alberto

1 - General information

Project code: PNRR-MR1-2022-12376596 **Project topic:** B1) Malattie rare: sostegno e potenziamento delle infrastrutture necessarie a sostenere la ricerca

PI / Coordinator: Burlina Alberto **Applicant Institution:** Veneto

Call section: Malattie Rare

Proposal title: A national model for dried blood spots biobanking, use for future research purposes and linking to National Registries of rare diseases

Duration in months: 24

MDC primary: Pediatria

000723

MDC secondary: Diagnostica

Project Classification IRG: Genes, Genomes and Genetics

Project Classification SS: Genetics of Health and Disease - GHD

Project Keyword 1: Discovery of genes and genetic variation for human health, disease, and disease susceptibility: Complex and Mendelian diseases such as psychiatric, neurological, ophthalmological, auditory, endocrinological, cardiovascular, developmental, reproductive, oncological, autoimmune, urological, respiratory; use of sophisticated genetic and genomic methods to identify candidate genes, single nucleotide polymorphisms, haplotypes, and copy number variation.

Project Request: **Animals:** ☐ **Humans:** ☒ **Clinical trial:** ☐

Project total financing request to the MOH: € 1.000.000

Free keywords: biobank, dried blood spot, newborn screening, gene expression profiling, storage.

Declarations

In case of a Synergy grant application 'Principal Investigator'(PI) means 'corresponding Principal Investigator on behalf of all Principal Investigators', and 'Host Institution' means 'corresponding Host Institution'.

1) The Principal Investigator declares to have the written consent of all participants on their participation and on the content of this proposal, as well as of any researcher mentioned in the proposal as participating in the project (either as other PI, team member or collaborator).	☒
2) The Principal Investigator declares that the information contained in this proposal is correct and complete.	☒
3) The Principal Investigator declares that all parts of this proposal comply with ethical principles (including the highest standards of research integrity — as set out, for instance, in the European Code of Conduct for Research Integrity — and including, in particular, avoiding fabrication, falsification, plagiarism or other research misconduct).	☒
4) The Principal Investigator is only responsible for the correctness of the information relating to his/her own organisation. Each applicant remains responsible for the correctness of the information related to him and declared above.	☒

The assessment of your grant application will involve the collection and processing of personal data (such as your name, address and CV), which will be performed pursuant to Regulation (EC) No 45/2001 on the protection of individuals with regard to the processing of personal data by the Community institutions and bodies and on the free movement of such data. Unless indicated otherwise, your replies to the questions in this form and any personal data requested are required to assess your grant application in accordance with the specifications of the call for proposals and will be processed solely for that purpose. Details concerning the purposes and means of the processing of your personal data as well as information on how to exercise your rights are available in the privacy statement. Applicants may lodge a complaint about the processing of their personal data with the European Data Protection Supervisor at any time.

Abstract

Newborn screening is a valuable public health prevention activity that continuously evolves to improve and optimize the health of our children.
One product of the multifaceted NBS process, the residual dried blood spots, serves as an additional valuable resource, whose benefits were discussed at length. Their overall use includes facilitating the improvement and evolution of NBS programs nationwide. It is envisioned that their uses will improve and change as scientific advances occur in the coming years.
Nowadays, the literature recognizes the three classes of residual RDBS use. These include (1) improvement of current screening programs; (2) introduction of new screening tests; and (3) expanding medical knowledge related to NBS.
Newborn screening is a non-invasive, risk-free test for the baby. It is a preventive medicine intervention allowing to identify some congenital diseases early, before the onset of symptoms. Early diagnosis enables to quickly start specific therapies to improve the child's health and prevent, in many cases, even serious and fatal complications. In Italy it is mandatory, free of charge and part of essential public health services.
In order to harmonize the regional situations, a nationwide screening program for inborn errors of metabolism was institutionalized by law between 2016 and 2017 (Law 167/2016,; DM 13 October 2016; DPCM 12-1-2017). The DM 13 October 2016 states that the screening program is a system articulated into four main functions (the screening laboratory, the laboratory for confirmatory diagnosis, the clinical centers, the regional coordination/supervision), defines the panel of screening conditions, the timing for specimen collection, the screening collection, the screening methodology, the confirmatory tests and the clinical follow up. A periodic review of the list of diseases being screened for, is set up by Ministry of Health, in collaboration with other government agencies and organizations (https://www.salute.gov.it/imgs/C_17_pagineAree_1920_0_file.pdf). No specific rules are reported about the use of residual DBS storage and the use after the screening for further investigations. Four Italian Neonatal screening Centers located in North (Padua), Centre (Florence) and South (Naples and Palermo) of Italy are currently identified by law as screening centers collecting more than 150,000/year DBS cardsw. The aim of our project will be to validate if dried blood samples are attractive for biobanking because of the ease and low cost of collection and storage.
The results of our study are encouraging as they suggest an inexpensive means to collect large numbers of blood samples, even by the donors themselves, and to transport, and store biobanked samples as spots of whole blood dried on paper. Combined with emerging means to measure hundreds or thousands of protein, such biobanks could prove of greatly enhancing discovery as well as routine analysis of biomarkers.
Our research will add information to standardize the following conditions and to establish guidelines for creating biobanks for collection of DBS for genetic purpose in Italy. we are moving to a future where biobanks existing biorepositories and reference database will be linked and networked for research purposes in way that has not been possible before.

In order to best review your application, do you agree that the above non-confidential proposal title and abstract can be used, without disclosing your identity, when contacting potential reviewers? Yes

2 - Participants & contacts

Project Code: PNRR-MR1-2022-12376596	**Call section:** Malattie Rare
Applicant Institution: Veneto	**Applicant/PI Coordinator:** Burlina Alberto

Operative Units

INSTITUTION	CF Institution	Department / Division / Laboratory	Role in the project	Southern Italy	SSN
1 - Veneto	00349040287	Division of Inherited Metabolic Diseases, Reference Centre Expanded Newborn Screening	Principal Investigator		X
2 - Università degli studi di Napoli Federico II	00876220633	Dipartimento di medicina molecolare e biotecnologie mediche	Research collaborator	X	X
3 - Firenze AOU Meyer	02175680483	Dipartimento Neuroscienze/Malattie Metaboliche/Lab Screening Neonatale	Research collaborator		X
4 - A.R.N.A.S. Civico e Di Cristina e Benefratelli/Sicilia	05841770828	Department of Pediatrics	Research collaborator	X	X

Principal Research Collaborators

Key Personnel Name	Operative Unit	Role in the project
1 - Salviati Leonardo	1 - Veneto	CoPI
2 - RUOPPOLO MARGHERITA	2 - Università degli studi di Napoli Federico II	Research collaborator
3 - Pavone Luigi Michele	2 - Università degli studi di Napoli Federico II	Research collaborator
4 - LA MARCA GIANCARLO	3 - Firenze AOU Meyer	Research collaborator
5 - CARDELLA FRANCESCA MARIA FATIMA	4 - A.R.N.A.S. Civico e Di Cristina e Benefratelli/Sicilia	Research collaborator
6 Under 40 - Gragnaniello Vincenza	1 - Veneto	Research collaborator
7 Under 40 - Gueraldi Daniela	1 - Veneto	Research collaborator

Key Personnel Name	Co-PI	Resp. CE	Resp. Animal	Birth Date	Gender
1 - Salviati Leonardo	X			15/11/1969	M
2 - RUOPPOLO MARGHERITA				08/05/1966	F
3 - Pavone Luigi Michele				17/04/1979	M
4 - LA MARCA GIANCARLO				22/02/1974	M
5 - CARDELLA FRANCESCA MARIA FATIMA				21/06/1955	F
6 Under 40 - Gragnaniello Vincenza				01/05/1988	F
7 Under 40 - Gueraldi Daniela				14/10/1986	F

Additional research collaborators under 40 to hire

Key Personnel Name	Operative Unit	Birth Date	Gender	Role in the project	Degree	Actual Pos. and Inst.
1 - PIROZZI FRANCESCA	2 - Università degli studi di Napoli Federico II	14/11/1995	F	Set up for bio bank	Medical Biotechnology	PhD student
2 - Scarcella Melania	2 - Università degli studi di Napoli Federico II	21/03/1996	F	Set up for bio bank	Medical Biotechnology	Research fellow
3 - PARTEXANO LAURA	4 - A.R.N.A.S. Civico e Di Cristina e Benefratelli/Sicilia	20/03/1991	F	biobank management and sample quality assessment	Biologist	unemployed

2.1 Administrative data of participating

Operative Unit Number 1:

Address: via Giustiniani,2 Padua 35128 Italy
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Address: via Pansini 5 Naples 80131 Italy
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Operative Unit Number 3:

Address: Viale Gaetano Pieraccini, 24, Florence 50139 Italy
PEC: meyer@postacert.toscana.it

Operative Unit Number 4:

Address: Piazza Nicola Leotta 4 Palermo 90127 Italy
PEC: ospedalecivicopa@pec.it

Operative Unit Number 5 (self financing):

Address: none
PEC: none