SIMPE CAMPANIA
SOCIETÀ ITALIANA MEDICI PEDIATRI

Carissimi,
è con enorme piacere che condivido con voi la soddisfazione di comunicarvi che l'abstract 'Targeted screening for the early identification of newborns with neuromuscular and neurometabolic disease' , accettato al World Summit on pediatrics 2023 che si terrà a Parigi a fine settembre.

Sono fermamente convinta che questa è la strada per accreditarci come società scientifica e cioè quella di analizzare e valutare l'enormità dei dati che produciamo quotidianamente, accompagnato da un saldo e forte rigore scientifico.

In allegato l'absctract

"Dear Dr.ssa Giuliano Maria,
We are pleased to inform you that you are officially invited to participate in this prestigious event which will be held in Paris, France.

The Wsp Paris 2023 Congress represents an important opportunity for doctors and medical professionals to share knowledge, experiences and innovations in the medical field.

Attached to this email, you will find the official invitation letter, which certifies the payment and your participation.

We kindly ask you to confirm your participation.

Furthermore, if there are specific requirements or special needs that you would like to report to us, we are at your disposal to provide assistance and support.

We await your participation in the Wsp Paris 2023 Congress with great interest and we hope to welcome you in Paris.

We remain at your disposal for further information or clarifications."

ABSTRACT

TARGETED SCREENING FOR THE EARLY IDENTIFICATION OF NEWBORNS WITH NEUROMUSCULAR AND NEUROMETABOLIC DISEASE

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INTRODUCTION
The phenotypic heterogeneity that characterizes the onset and development of some neurometabolic and neuromuscular disorders can be reason for a diagnostic delay. Aromatic amino acid dopa decarboxylase (AADCD) deficiency and Duchenne muscular dystrophy (DMD) are two conditions for which there are more effective treatments if diagnosed early.
Our study aimed at the early identification of AADCD and DMD deficiency through the application of biomarkers 3-O-Methyldopa (3-OMD) and CK-MM on Dried Blood Spots (DBS), performed in the medical clinic of family pediatricians (PdF) in the Campania and Lazio regions, in collaboration with the Child Neuropsychiatry of the Sapienza University (AOU Policlinico Umberto I of Rome) and the regional reference laboratory for newborn screening of the Lazio region (LRR).
METHODS
After collecting the informed consents, DBS samples were gathered by pediatricians, from both male newborns and newborns showing clinical manifestations indicative of the two pathologies that were subjects of the study. Latier hypotonia, oculogyric crisis, dystonia, and neuromotor developmental delay samples were sent to the LRR for analysis. The dosage of 3-OMD was collected by using UPLC-ESI- MS/MS, as previously specified (De Carlo et al.), followed by an CK-MM immunofluorimetry assay on GSP ® platform. Screened positive subjects underwent molecular confirmation by direct sequencing for the DDC gene and MLPA analysis of the DMD gene.
RESULTS
A total of 290 DBS were analysed, 80 for the 3-OMD assay and 267 for CK-MM. For the cases with borderline values, direct sequencing molecular testing of the DDC gene for AADCD and MLPA analysis for DMD were performed. No positive cases were identified.
DISCUSSION AND CONCLUSION
Targeted screening for AADCD and DMD is an important strategy for the early identification of rare and treatable diseases. The use of DBS in PdF clinics facilitates the execution of the screening test on a large number of subjects, in a short time period, and does not require the support of hospital structures. The methodological approach reported in this study can be adapted and implemented in different territorial contexts, allowing for the early identification of these pathological conditions during the asymptomatic phase and opening the way for novel pharmacological strategies before the complete development of the pathology.
ACKNOWLEDGMENTS: we thank the PdF of Campania (Antignani R., Brunese F.,De Prosperis A., Del Gaizo D., Opallo A., Sassi R., Stabile D.) and Lazio (Castronuovo S., Giannantonio A., Grimaldi V., Labella I., Morano D., Vitolo R., Zirletta E.) for their precious support in collecting DBS samples.