CRML

Site constitutif CHU de Rouen

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Bekri et al. 2021 (soumis)

Centre de Référence de Maladies Lysosomales – Site CHU de Rouen

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Equipe Clinique			Equipe Biologique		
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Médecine Interne	Dr G. SAUVETRE	CHU de Rouen			
Génétique clinique	Dr AM. GUERROT Dr A. GOLDENBERG	CHU de Rouen			
Centre Greffe Moëlle Osseuse Pédiatrique	Pr P. SCHNEIDER, Dr N. BUCHBINDER	CHU de Rouen			

Analyse Intégrée Multimodales en Santé

AIMS

File active
110 patients

Projets en cours

MetaLys	Caractérisation multi-omique des maladies lysosomales (**>30 publications**)
LysoNeo	Dépistage de 13 maladies lysosomales en Haute Normandie (~100,000 patients inclus)

Partenariat - Recherche

INSERM U1088, Pr. Jérome AUSSEIL, Université de Toulouse

CNRS UMR6014, Pr. Carlos AFONSO, Université de Rouen Normandie

AIMS, Dr Olivier Lidove, Wladimir Mauhin, centre de référence des maladies lysosomales, Service de médecine interne, Groupe Hospitalier Diaconesses-Croix Saint Simon,

EA7453, Pr. Marc Berger Hématologie Biologique, CHU Clermont-Ferrand

LysoNeo Project

Réseau Périnatalité Normandie

LysoNeo - Investigators

Promotor	**Rouen University Hospital**
Ethical approval	**May 2020**
Start	**March 2021**
Duration	**3 years**
Inclusions	**100,000**

Réseau de Périnatalité de Normandie

Coordinator	**Prof.**	**Soumeya**	**BEKRI**	**Metabolic Biochemistry Department - LBM Rouen University Hospital**
Principal Investigator	**Dr.**	**David**	**GUENET**	**Biochemistry Department, CRDN Normandie (Regional Newborn Screening Center), Caen University Hospital**
Principal Investigator	**Prof.**	**Stéphane**	**MARRET**	**Department of Neonatal Pediatrics, Intensive Care and Neuropediatrics, Rouen University Hospital**
Investigateurs				**Normandy Perinatal Network 23 Maternity Hospitals**

Public-Private Partnership

Objectives

Primary Objective

To evaluate the feasibility of systematic neonatal screening for **MPS I** and **Pompe disease** by multiplex analysis of lysosomal enzyme activities using Dried Blood Spots in a cohort of 100,000 newborns in the Normandy region.

Ancillary study

Screening for 9 other lysosomal pathologies: Krabbe disease, Niemann-Pick A/B disease, mucopolysaccharidoses (MPSII, MPSIIIB, MPSIVA, MPSVIB, MPSVI and MPSVII), neuronal lipofuscinosis type 2 (CLN2).

Lysosomal Acid Lipase Deficiency

Metachromatic Leucodystrophy

Promotor	**Rouen University Hospital**
Ethical approval	**January 2022**
Start	**March 2022**
Time Period	**2 years**
Inclusions	**60,000**

Inclusions

Inclusion criteria

Newborns born in a Maternity Hospital in Normandy

Newborns participating in the National Neonatal Screening Program

Signed consent form

Non-inclusion criteria

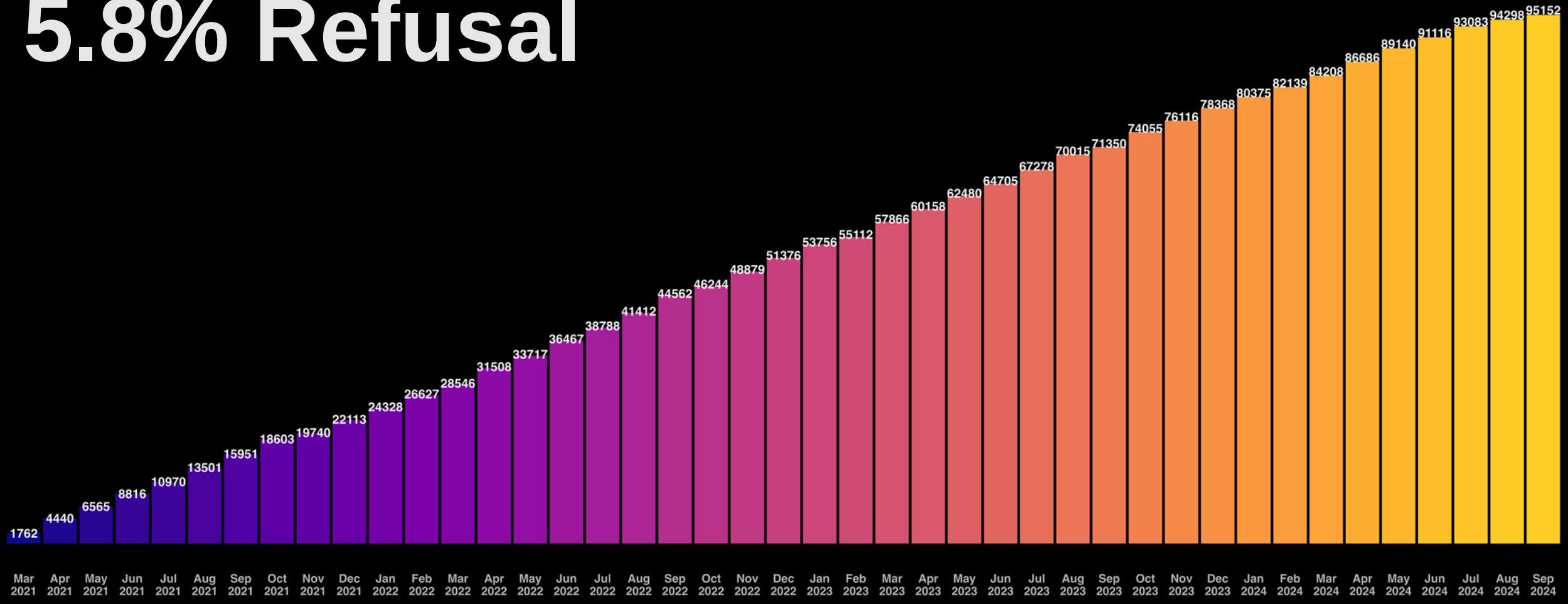
There are no criteria for non-inclusion

Tiered Investigations

	1st Tier	2nd Tier		
MPS	Enzyme	GAG	GAG (Urine)	Genotyping
Pompe	Enzyme	Glc4 (Urine)		Genotyping
Krabbe	Enzyme	Psychosine		Genotyping
NPA/B	Enzyme	LysoSM		Genotyping
MLD	Sulfatides	Enzyme		Genotyping
CLN2	Enzyme			Genotyping
LALD	Enzyme			Genotyping

LysoNeo Primary Study Update Sep 2024

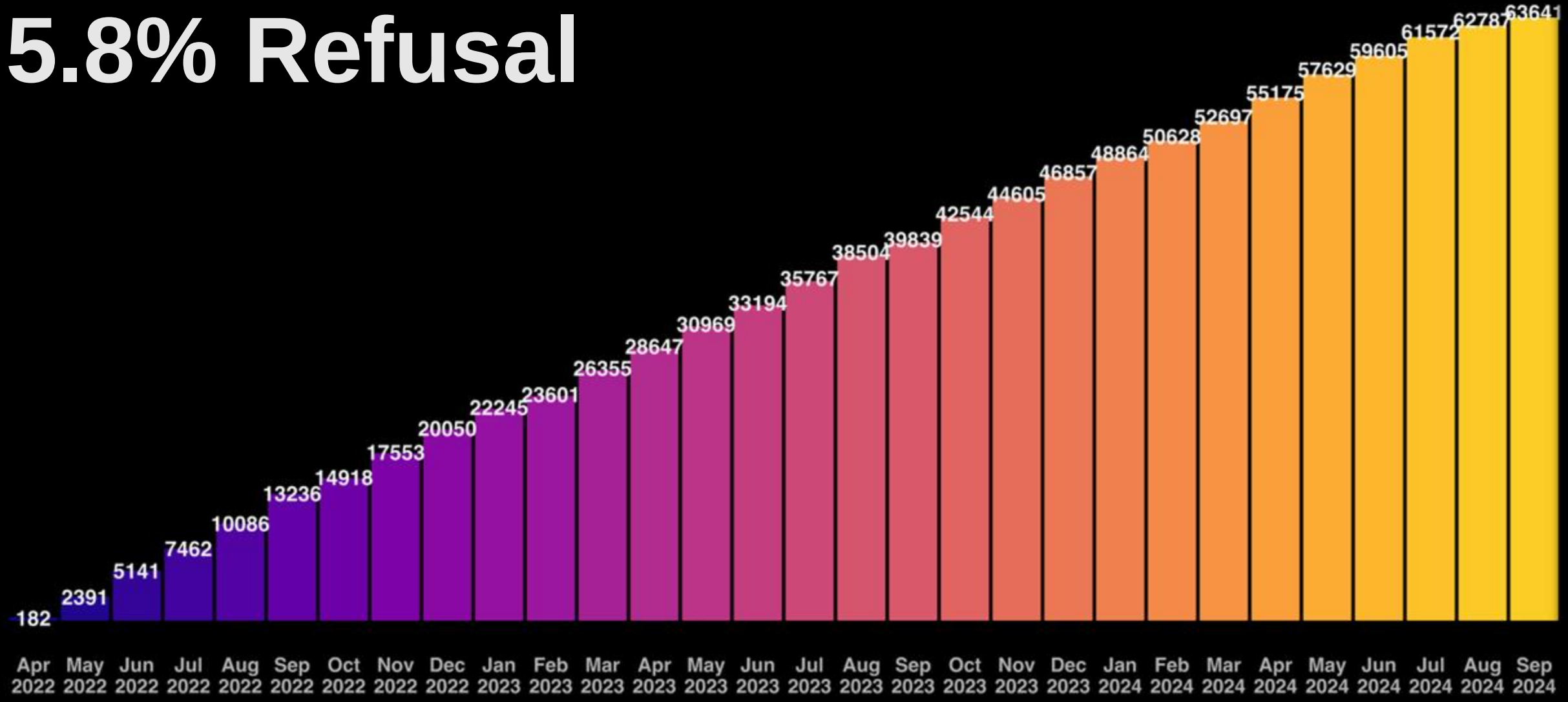
5.8% Refusal



Unpublished data - CHU de Rouen (LBM)

LysoNeo Extension Study Update Sep 2024

5.8% Refusal



Unpublished data - CHU de Rouen (LBM)

Perspectives

June 2024	HAS: extension of national NBS program to MPS1, Pompe disease, MLD and acid lipase deficiency
July 2024	Referral to HAS by patient association ELA for MLD NBS
July 2024	HAS self-referral for LD NBS
November 2004	LysoNeo report to be addressed to HAS

LysoNeo Project Extension - 2024 - 2026

Metachromatic Leucodystrophy

Lysosomal Acid Lipase Deficiency

Pompe disease

Mucopolysaccharidosis type 1

Promotor	**Rouen University Hospital**
Ethical approval	**October 2024**
Start	**November 2024**
Duration	**2 years**
Inclusions	**60,000 newborns**

LysoNeo Project Extension - 2024 - 2026

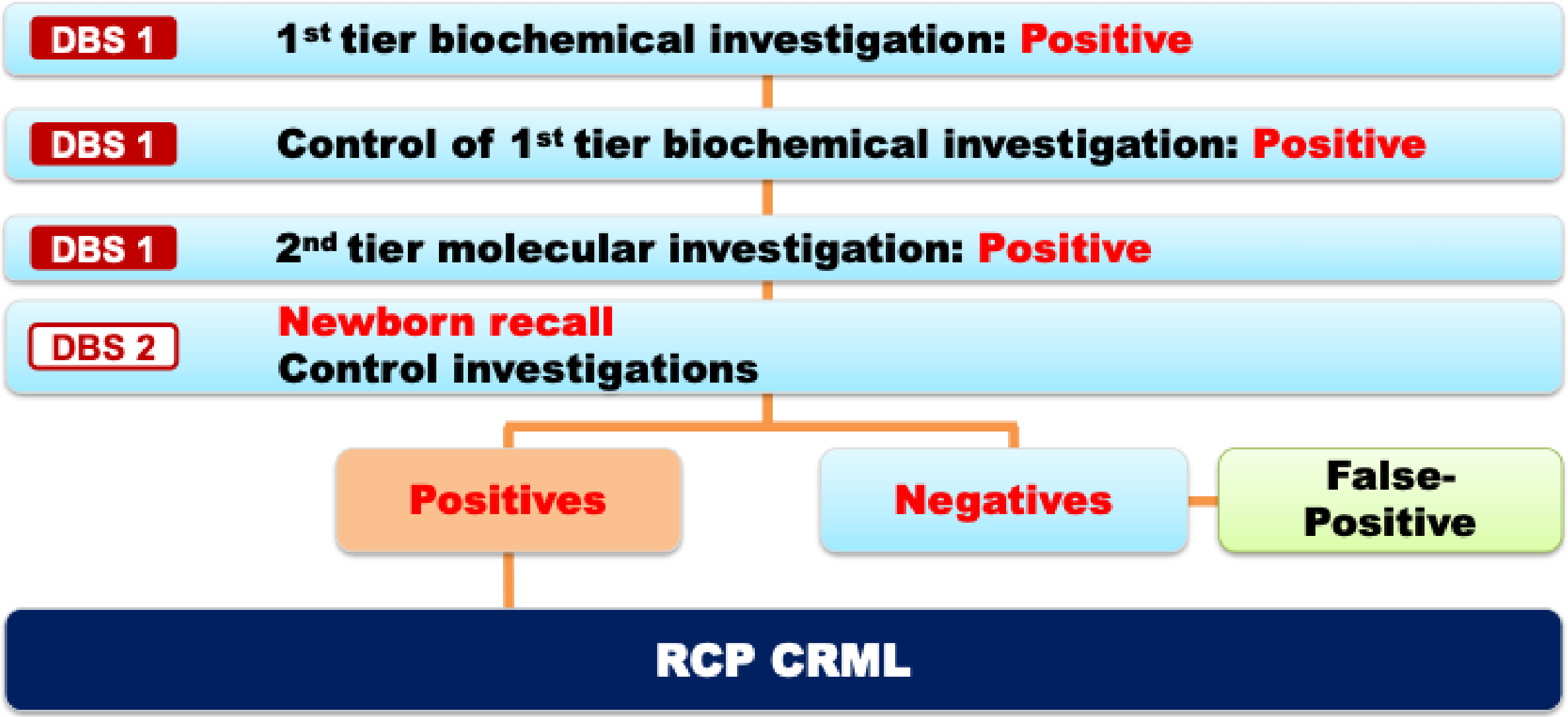
1st tier biochemical investigations → False positives

Unaffected newborns were called

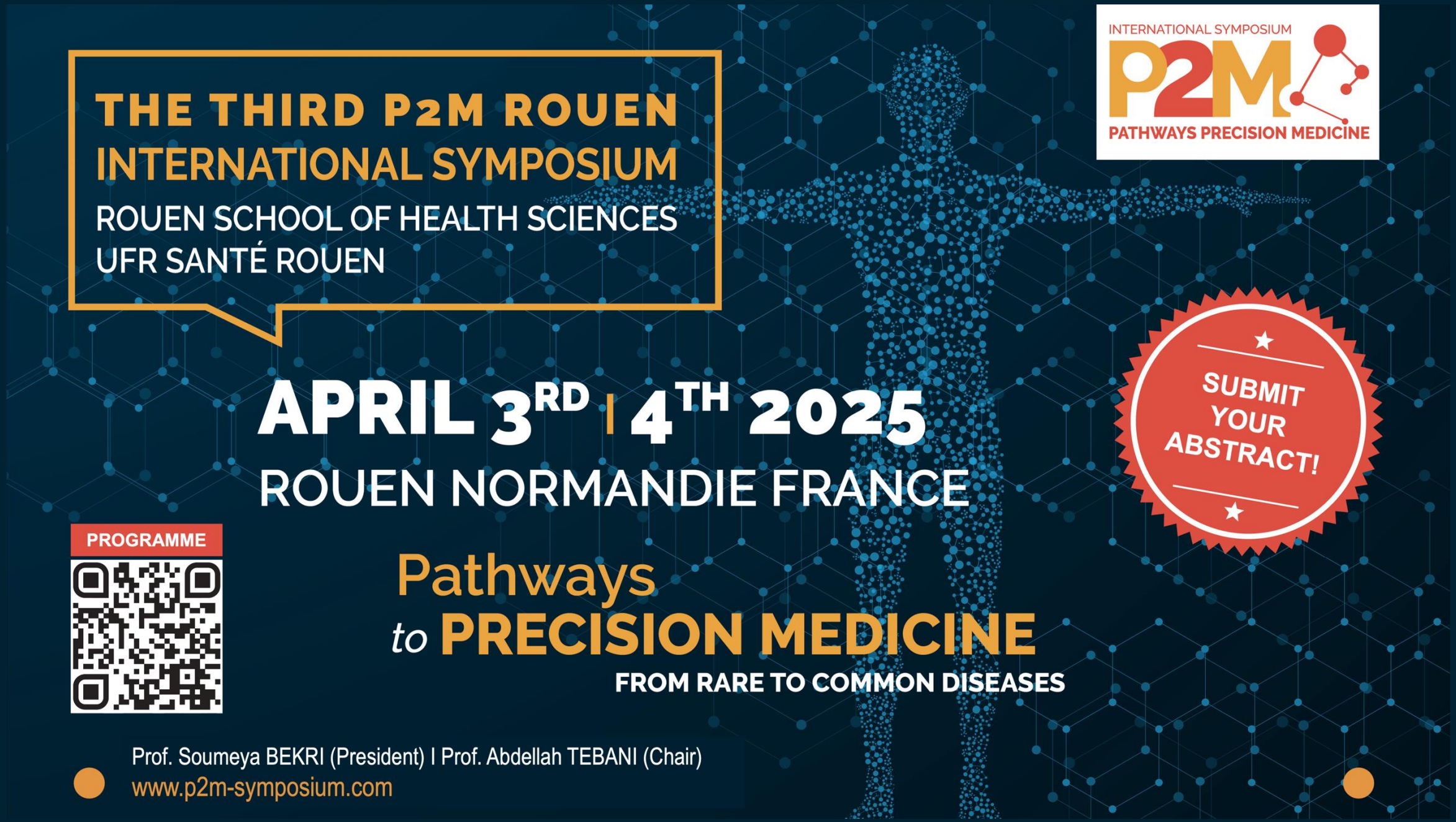
Parental stress

Unnecessary burden on caregivers

LysoNeo Project Extension - 2024 - 2026



CPP Approval October 2024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Prof. Soumeya BEKRI (President) I Prof. Abdellah TEBANI (Chair)
www.p2m-symposium.com

Acknowledgements

Participants: newborns and parents
Medical Staff
CRML
Partners

LysoNeo

Systematic Neonatal Screening for Lysosomal Diseases: French Pilot Study

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