Evolution and geographical patterns of the Portuguese Newborn Screening program in the last 10 years

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OBJECTIVES:

The Portuguese National Neonatal Screening Program (PNRN) covers 28 diseases and successfully analyses more than 99% of the newborns in the country. It allows the early diagnosis and treatment of hereditary metabolic diseases and spinal muscular atrophy, among other highly debilitating diseases, responsible for significant morbidity and mortality. Early diagnosis can contribute to better patients' outcomes and improved patients' quality of life.
This study aimed to investigate the evolution and geographical distribution of the diseases included in the panel of diseases of the PNRN from 2013 to 2023.

METHODS:

The PNRN started in 1979 with phenylketonuria (PKU) screening, an inherited metabolic disorder. Since then, it has evolved to include congenital hypothyroidism (CH), other inherited metabolic disorders (IMD), cystic fibrosis (CF), sickle cell disease (SCD) and spinal muscular atrophy (SMA). Data was collected from the last 10 yearly reports of PNRN publicly available [1].
Summary measures of disease incidence were analysed, and chronologically and geographically reported. The incidence of the diseases included in this study was calculated in cases per 100,000 (one hundred thousand) inhabitants.

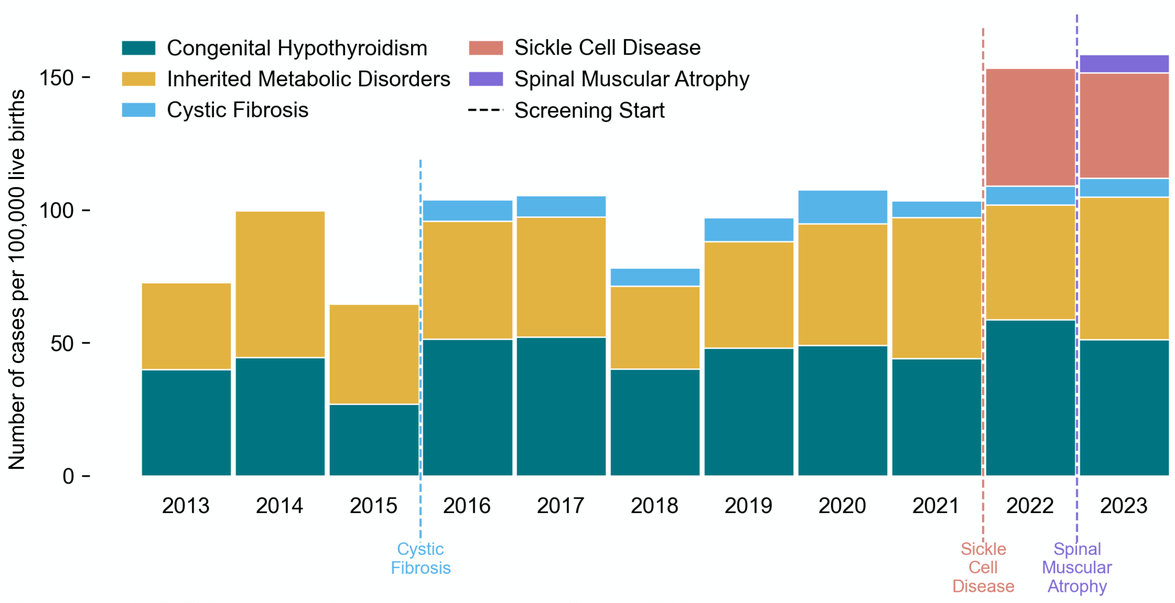
RESULTS:

In total, 971 cases of rare diseases were diagnosed through PNRN, representing a two-fold increase between 2013 and 2023. This increment was 54% due to incidence increment and 46% due to the addition of SCD and SMA into the panel of screened diseases. Of the 971 diagnosed cases of rare diseases, there were 12 with unknown geographical location: 2 cases of IMD (2014), 1 case of IMD and 8 cases of CF (2019) and 1 case of SCD (2022).

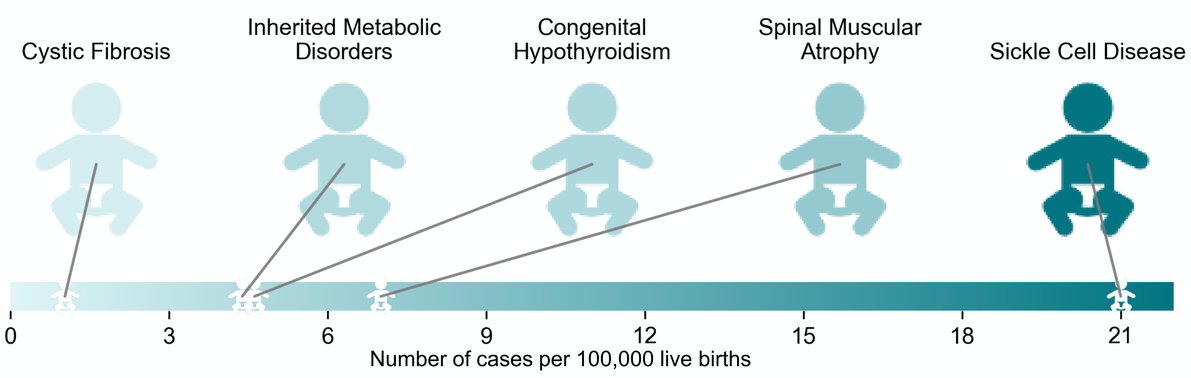
The 10-year incidence rate was 4.4 and 4.6 cases per 100,000 screens for IMD and CH, respectively. These incidence rates present an increasing trend since 2013.

Additionally, the 8-year incidence rate was 1.0 case per 100,000 screens for CF. The 2-year incidence rate was 21.0 cases per 100,000 screens for SCD. The 1-year incidence rate was 7.0 cases per 100,000 screens for SMA.

Evolution of Neonatal Disease Incidence



Neonatal Disease Incidence by Condition



RESULTS (CONTINUATION):

The highest incidence of screened rare diseases was found in the south region of Portugal (Beja).

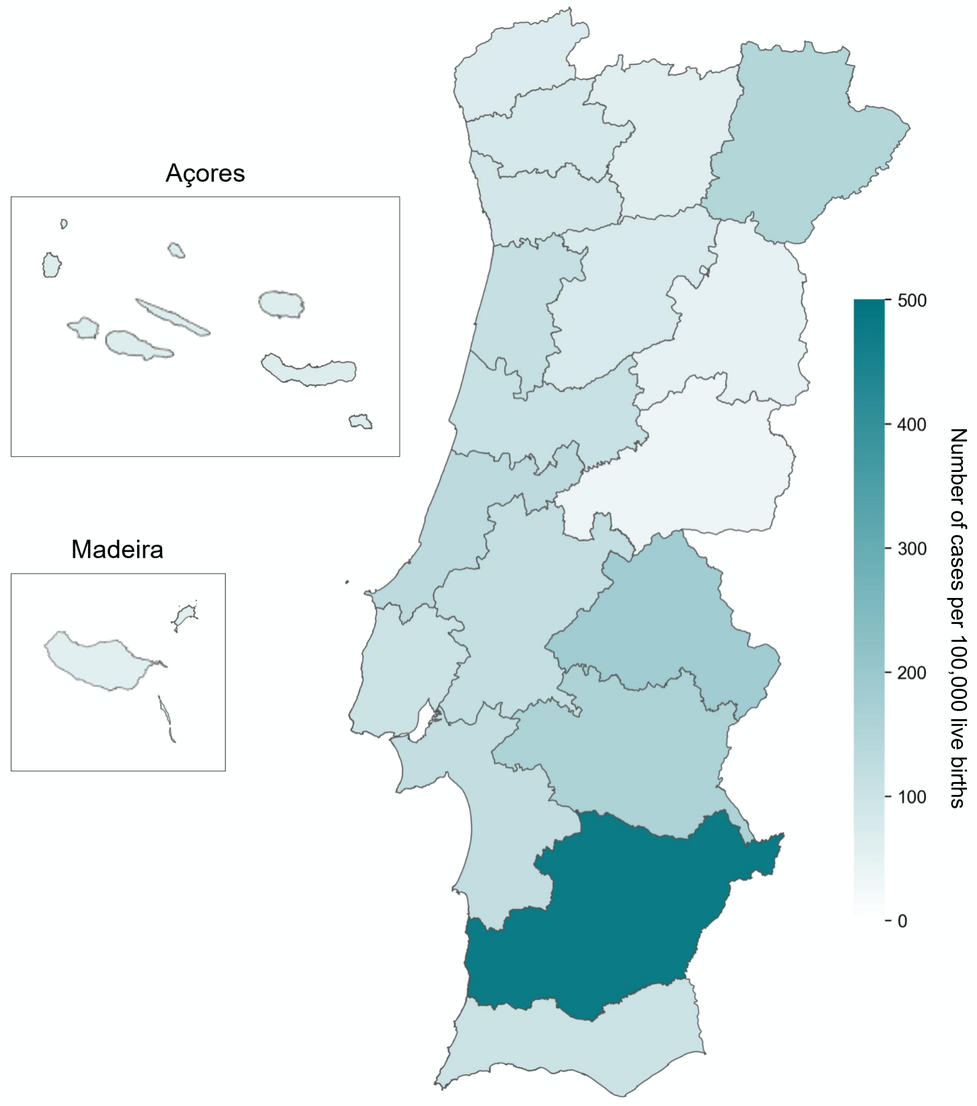
Beja, with a total incidence of 477.4 cases of rare diseases per 100,000 screens, was followed by Portalegre, with 184.8 cases of rare diseases per 100,000 screens, and then by Évora, with 161.3 cases of rare diseases per 100,000 screens.

The lowest incidence of screened rare diseases was found in the central region of Portugal (Castelo Branco).

Castelo Branco, with a total incidence of 36.1 cases of rare diseases per 100,000 screens, was followed by Guarda, with 50.6 cases of rare diseases per 100,000 screens and then by the Autonomous Region of Madeira, with 60.0 cases of rare diseases per 100,000 screens.

The remainder of the country revealed a homogeneous distribution of cases.

Neonatal Disease Incidence by District



= CONCLUSIONS:

Nationwide newborn screening programs are essential to understand the epidemiology of rare diseases. In Portugal newborn screening diseases are increasing in its incidence with a relatively homogeneous distribution across the country.

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