RARE DISEASE PERSON'S CARD

Technical Report

2024

REPÚBLICA PORTUGUESA
SAÚDE
DGS Direção-Geral da Saúde

RARE DISEASE PERSON'S CARD

Technical Report

TECHNICAL FILE
Portugal. Ministry of Health. Directorate-General of Health.
TECHNICAL REPORT
RARE DISEASE PERSON'S CARD
Lisbon: Directorate-General of Health, 2025.

KEYWORDS
RARE DISEASE PERSON'S CARD

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Lisbon, November 2025

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Resumo

O que é este documento?

O relatório anual sobre a implementação do Cartão da Pessoa com Doença Rara (CPDR), referente ao ano de 2024.

O que consta do documento?

Neste relatório anual são apresentados dados que demonstram a implementação do processo de requisição do CPDR no ano de 2024.

Quais são as principais conclusões?

Em cumprimento da Norma da DGS n.º 01/2018, verificou-se que durante o ano de 2024 foram emitidos 1593 CPDR, podendo observar-se o registo de 515 doenças raras diferentes nos novos cartões emitidos, das quais 117 são novas doenças codificadas pela primeira vez no ano de 2024. Trinta e quatro unidades de consultas de especialidade médica emitiram CPDR neste ano.

O que se quer atingir em 2025?

- Iniciar o processo de visualização do CPDR nos sistemas de informação das urgências dos hospitais no momento da triagem;
- Aumentar e atualizar os códigos ORPHA disponíveis no CPDR.

Summary

What is this document?

The 2024 Annual Report on the implementation of the Rare Disease Person's Card (RDPC) has been prepared by the Department of Quality in Health, Division of Quality Design and Improvement.

What can I find in this document?

This annual report presents data regarding the implementation process of the RDPC request during the year 2024.

What are the main conclusions?

In compliance with the DGS Guideline No. 01/2018, during 2024, 1,593 RDPCs were issued, and 515 different rare diseases were registered, including 117 diseases registered for the first time in 2024. This year thirty-four specialized medical units have registered RDPCs.

What do we aim for 2025?

- Start the visualization of the RDPC in the existing information systems of the emergency departments during the triage;
- Increase and update the available ORPHA codes in the RDPCs.

Introduction

The European definition of a rare disease, adopted by the Directorate-General of Health (DGS), corresponds to diseases with a prevalence of no more than 5:10,000 (European Commission, 2014)[1].

Rare diseases exhibit the following characteristics:

a) They are chronic diseases, many of them severe and degenerative, often hereditary;

b) They can manifest in any age group;

c) They present a wide diversity of symptoms and signs that vary not only from disease to disease but also from patient to patient suffering from the same disease;

d) They can be progressively disabling and may negatively impact quality of life and life expectancy;

e) There is no cure for most of them;

f) They may cause significant suffering for patients and their families;

g) They can be associated with a deficit of medical and scientific knowledge, due to their rarity;

h) Medical care can improve the quality of life of those affected and extend their life expectancy.

The variability in approach, treatment, and clinical monitoring, especially in urgent and emergency situations, justifies the need for establishing a special protection mechanism for people living with a rare disease. As recognition of this need, the Portuguese Parliament Resolution No. 34/2009 was approved and published in Diário da República, 1st Series, No. 88, of 7 May, to promote the creation of a "Card" for people living with a rare disease.

In 2014, following these policies, the Directorate-General of Health (DGS), through the Department of Quality in Health, developed an instrument for the special protection of people living with a rare disease, titled the 'Rare Disease Person's Card' (RDPC) – in Portuguese, "Cartão da Pessoa com Doença Rara (CPDR)", with the following aims:

a) To ensure that healthcare professionals can access relevant information regarding people living with a rare disease and their clinical data, enabling better health care for these patients in emergent and/or urgent situations.

b) To improve continuity of care by ensuring patient access to all their relevant clinical information in an accessible format that accompanies them across different levels of healthcare.

c) To enable the prompt appropriate clinical referral to the healthcare unit that effectively provides the proper healthcare for the patient.

[1] European Commission (2014). Report on the Implementation of the Commission Communication on Rare Diseases: Challenges for Europe [COM (2008) 679 final] and the Council Recommendation of 8 June 2009 on an Action in the Field of Rare Diseases (2009/C 151/02). Available at: https://eur-lex.europa.eu/legal-content/PT/TXT/PDF/?uri=CELEX:52014DC0548&from=PT [Accessed on 30 June 2020].

This report outlines the progress of the RDPC implementation process, with a focus on the monitoring data for the year 2024. To streamline the process and clarify the RDPC issuance procedure, DGS Standard No. 01/2018 defines its issuance and consultation conditions.

The RDPC request is made by the attending physician through the Electronic Health Record Platform, which provides, within the professional interface, a list of rare diseases along with their corresponding ORPHA code and specific healthcare considerations for emergency and urgent care contexts. These considerations are editable by the physician, allowing for the customization and adjustment of information to the specific case of the individual with a rare disease, ensuring personalized emergency and urgent care.

Advancements in the use of the RDPC not only highlight the DGS's commitment to protecting individuals with rare diseases but also position Portugal as a reference in the European context for the treatment and management of these conditions. This ongoing effort ensures:

a) Improved continuity of care, with simplified access to clinical information in emergency services;

b) Personalization of emergency treatments, based on ORPHA codes and specific guidelines;

c) Increased early and accurate diagnosis, due to the integration of new codes and the continuous training of healthcare professionals.

Results of the RDCP Implementation Process

Since 2014, as observed in **Table 1**, healthcare professionals and units have shown increasing interest in requesting RDPCs, with 13,263 RDPCs having been requested up to December 31, 2024.

Table 1. Rare Disease Person's Card Implementation

INDICATOR	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Number of cards requested	622	911	776	2703	1100	1065	895	1142	1039	1417	1593
Number of issuing healthcare units	6	13	14	24	30	25	24	25	30	35	34
Number of new diseases coded	168	182	106	225	150	145	90	97	82	110	117

The number of new RDPCs issued in 2024 increased by 12.4% compared to 2023, which could be related to the training sessions conducted for physicians and facilitated by the Directorate-General of Health (DGS), within the scope of ORPHA codification and the European Project OD4RD (Orphanet Data For Rare Diseases). Otherwise, 2017 data indicate a significant increase in the number of cards requested, largely related to the expanded possibility of requesting RDPCs to all public and private hospitals, which became responsible for its implementation and dynamization. However, a stabilization of the number of issued cards each year is expected due to various factors such as:

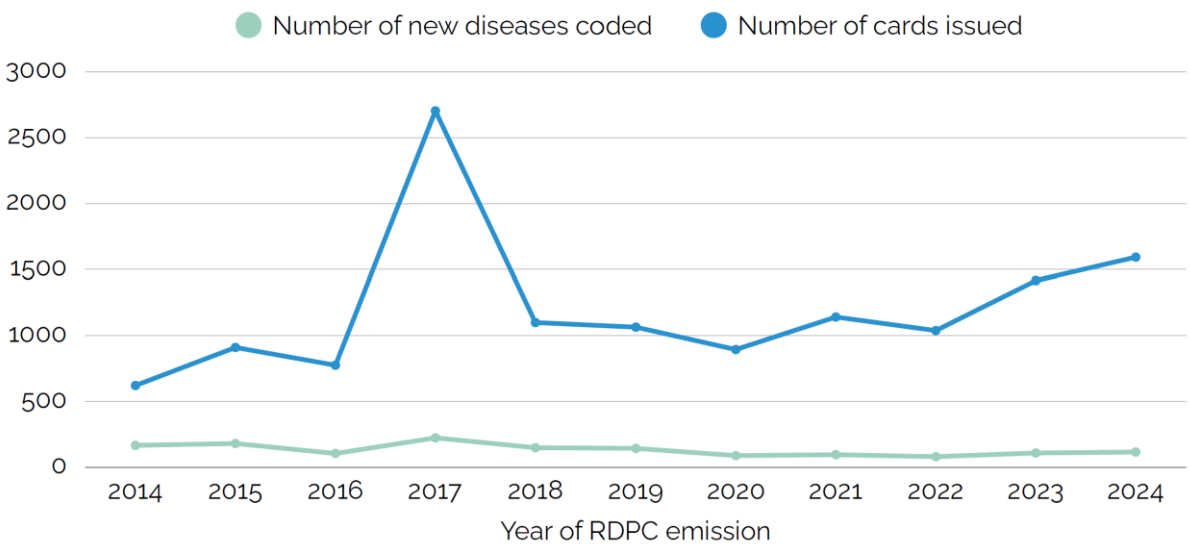
a) Obtaining a rare disease diagnosis is mostly a complex and lengthy process, reason why diagnoses are usually only made by Reference Centres.

b) An increase in the level of public awareness around the General Data Protection Regulation may have an impact on the number of RDPCs issued, as it is dependent on written consent.

c) Recently constraints in updating the available ORPHA codes in RDPC.

However, comparing the number of new diseases coded with ORPHA codes in 2024 to the total of diseases coded in previous years, it is possible to conclude that 117 new rare diseases were coded for the first time in 2024 (**Graph 1**).

Graph 1. Evolution of the Implementation of Rare Disease Person's Card



By analysing individual information by healthcare provider, it can be observed that, in 2024, the 1,593 RDPCs requested were issued by 34 institutions, as shown in **Graph 2**, one fewer than in 2023. It was also verified that 82.7% of RDPCs were requested by seven Local Health Units (ULS) and by the Portuguese Institute of Oncology (IPO) of Porto and IPO of Lisbon, which include Reference Centres for rare diseases, namely:

a) *Unidade Local de Saúde de Coimbra, E.P.E. (32,8%);*

b) *Unidade Local de Saúde de São José, E.P.E. (15,4%),* of which *14,0%* were requested by *Centro Hospitalar Universitário de Lisboa Central* and *1,4%* by *Instituto de Oftalmologia Dr. Gama Pinto;*

c) *Unidade Local de Saúde de Santo António, E.P.E. (12,1%);*

d) *Unidade Local de Saúde de Santa Maria, E.P.E. (11,9%);*

e) *Unidade Local de Saúde de São João, E.P.E. (8,5%);*

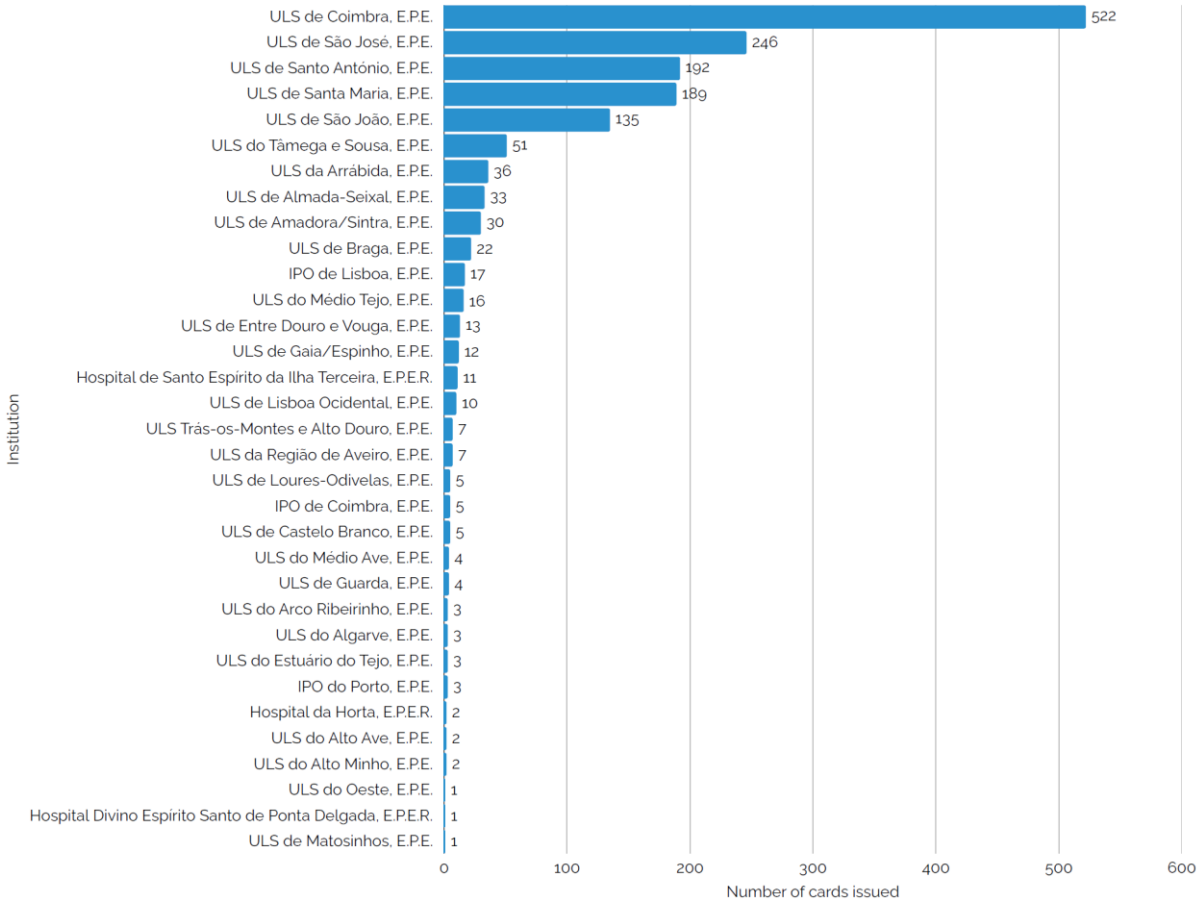
f) *Instituto Português de Oncologia de Lisboa Francisco Gentil, E.P.E. (1,1%);*

g) *Unidade Local de Saúde de Lisboa Ocidental, E.P.E. (0,6%);*

h) *Instituto Português de Oncologia do Porto Francisco Gentil, E.P.E. (0,2%);*

i) *Unidade Local de Saúde do Alto Ave, E.P.E. (0,1%).*

Graph 2. Number of RDPCs requested by Institution in 2024



Analysing the information by disease, it can be noted that in 2024 a total of 515 different rare diseases (515 distinct ORPHA codes) were recorded in the newly issued cards — corresponding to around 7–8% of the universe of rare diseases described in the Orphanet database. Of these, 117 represent new diseases coded for the first time in 2024.

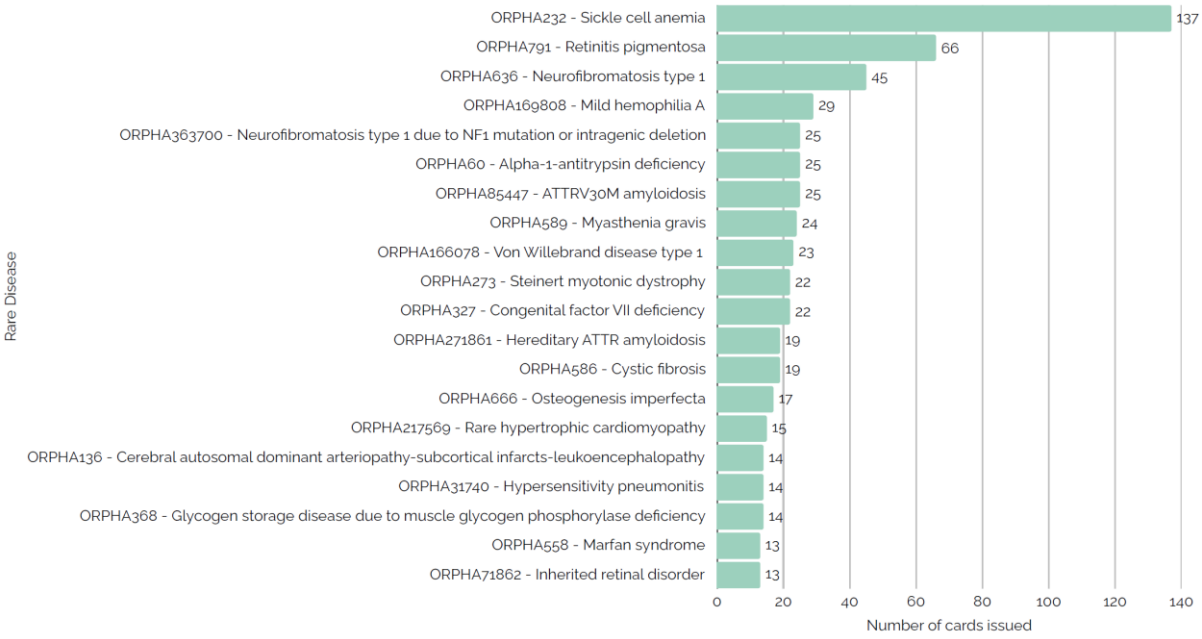
As in 2023, the rare diseases with the highest number of RDPC requests in 2024 were Sickle Cell Anemia, Retinitis Pigmentosa and Neurofibromatosis type 1 (**Graph 3**). These diseases are also among the most prevalent in Europe, according to the 2024 list published annually by Orphanet[2].

In 2024, the 1,593 cards issued corresponded to 1,592 individuals living with a rare disease. Over time, the issuance of a new card for the same person may occasionally occur. This may be associated either with the update of a preliminary diagnosis to a more precise one—corresponding to an updated designation of the rare disease—or with the identification of a distinct rare disease.

Between 2014, the year RDPC issuance began in Portugal, and the end of 2024, a total of 1,472 different rare diseases were identified (**cf. Table 1**), corresponding to 11,834 individuals with a rare disease holding an RDPC. Currently, 6,528 rare disease codes are described on the Orphanet website.

[2] Orphanet Report Series (2024). Prevalence of Rare Diseases: Bibliographic Data – Number 2. Available at: https://www.orpha.net/pdfs/orphacom/cahiers/docs/PT/Prevalencia_das_doencas_raras_por_prevalencia_decrescente_ou_casos.pdf [Accessed on 14 April 2025].

Graph 3. Rare diseases with the most cards requested in 2024



To provide a more detailed characterization of rare diseases recorded in RDPC issuance, cards issued in 2024 were grouped according to the Orphanet Classification, as shown in **Graph 4**. This categorization was based on the correspondence between the ORPHA codes of the cards and the corresponding nosological classification. It should be noted that, in this analysis, the total number of RDPCs by Orphanet classification (1,601) exceeds the total number of cards actually issued (1,593 in 2024). This discrepancy arises from the fact that a single card may be associated with more than one diagnosis, reflecting the possible coexistence of multiple rare diseases in the same individual. Thus, each RDPC may contribute to more than one diagnostic category, which explains the difference between the total number of cards and the sum of records by disease group. Additionally, some cards have ORPHA codes that do not fall under any classification due to recurring updates in the ORPHA nomenclature. In such cases, they are categorized under Orphanet-defined concepts such as "obsolete entity"—when the disease is better described by a more precise term or by an existing, more detailed classification—or "not rare in Europe."

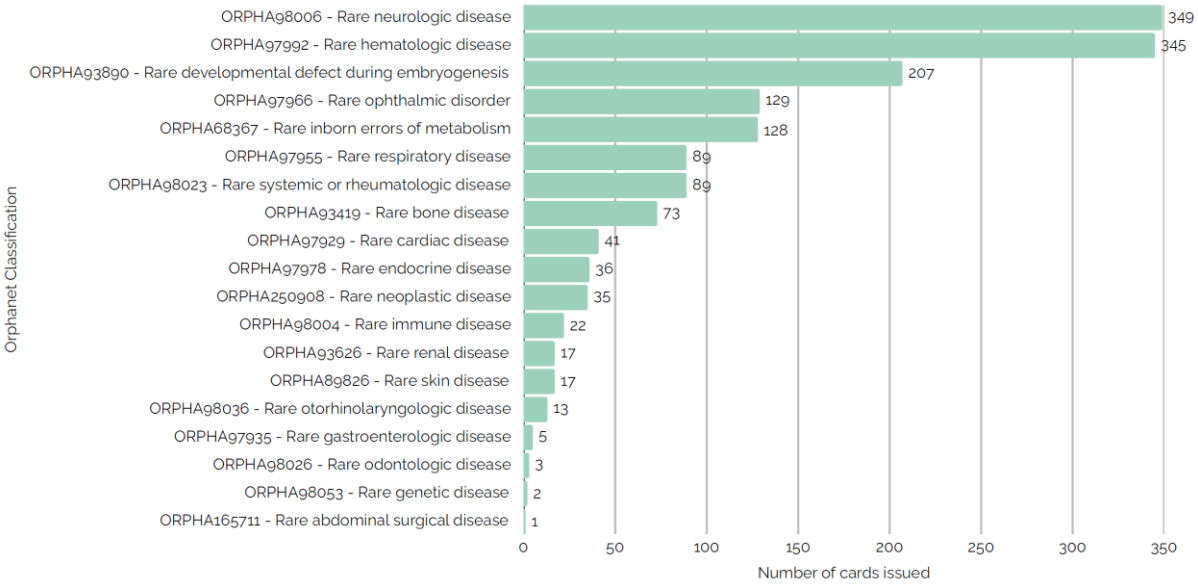
In 2024, the disease groups with the highest number of RDPCs issued were rare hematological diseases (e.g., Sickle Cell Anemia), rare neurological diseases (e.g., Neurofibromatosis Type 1), and rare ophthalmological diseases (e.g., Retinitis Pigmentosa). This epidemiological profile aligns with European prevalence data provided by Orphanet through Orphadata (December 2024 version)[3].

It is noteworthy that some of these diseases—such as Sickle Cell Anemia, Cystic Fibrosis, and Phenylketonuria—are included in the national neonatal screening panel, commonly referred to as the "heel-prick test." (in Portuguese, "teste do pezinho"[4]. This public health program, promoted by the National Institute of Health Dr. Ricardo Jorge (INSA), enables early detection of several rare metabolic and hematological diseases, significantly increasing the likelihood of diagnosis within the first days of life and, consequently, the timely issuance of an RDPC.

[3] Orphanet. Knowledge Base Release, December 2024. Epidemiology (prevalence and/or incidence) of Orphanet rare diseases. Available at: https://www.orphadata.com/epidemiology/ [Accessed on 18 June 2025]

[4] National Health Service (2025). Neonatal Screening. Available at: https://www.sns24.gov.pt/pt/tema/saude-da-crianca/rastreio-neonatal/. [Accessed on 10 June 2025]

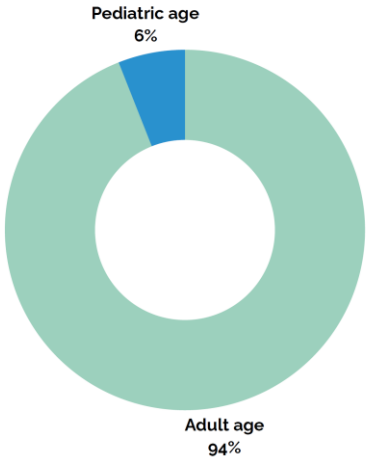
Graph 4. Number of RDPCs issued in 2024, by Group of Diseases according to the Orphanet Classification



An exploratory analysis of RDPC issuance in the pediatric age group was conducted based on the issuing ULS. "Pediatric RDPCs" were defined as cards issued by services specialized in pediatric care, including Pediatrics, Pediatric Neurology, Pediatric Genetics, Pediatric Hematology, Pediatric Endocrinology, Pediatric Cardiology, Pediatric Pulmonology, and the Luís Borges Child Development Center. Given the absence of age or date-of-birth variables in the RDPC database, this approach served as an indirect indicator of age group. While exceptions may occur—such as adult patients remaining in pediatric services due to chronic or rare conditions—this method provides a reasonable proxy for preliminary age-group segmentation.

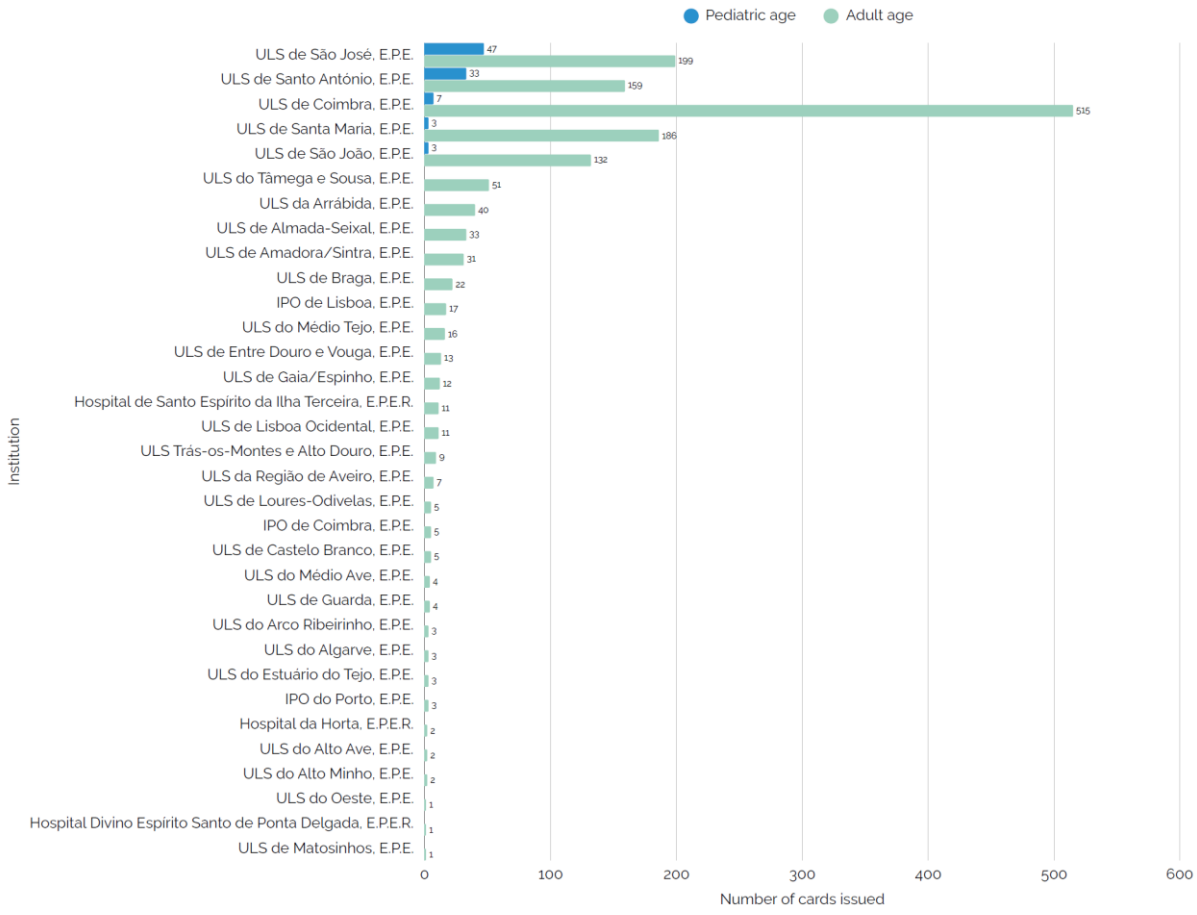
In 2024, 93 RDPCs were issued by these services, accounting for approximately 6% of all cards issued that year (**Graph 5**).

Graph 5. Proportion of RDPCs issued in the pediatric age group in 2024



As shown in **Graph 6**, the issuance of RDPCs in the pediatric age group was concentrated exclusively in institutions that are part of the National Reference Centers. This pattern contrasts with RDPC issuance in adults, which aligns with the global trend observed in 2024 (**see Graph 2**) and is characterized by a greater diversity of requesting institutions, reflecting a broader distribution.

Graph 6. Number of RDPCs issued to pediatric and adult patients in 2024, by institution



This contrast highlights the importance of strengthening the standardization of practices and continuous training to ensure equity and consistency in RDPC issuance across the National Health Service (SNS).

The higher proportion of certain rare diseases among RDPCs issued in the pediatric age group in 2024—including Sickle Cell Anemia and Cystic Fibrosis (**Tables 2 and 3**)—can be largely explained by the National Neonatal Screening Program (PNRN), commonly referred to as the "heel-prick test," as previously noted.

In 2024, this program screened 84,631 newborns in Portugal, identifying 133 positive cases of rare diseases. Among these, there were 51 inherited metabolic disorders, 36 cases of sickle cell disease (clinical form of Sickle Cell Anemia), and 6 cases of Cystic Fibrosis. In the same year, 30 RDPCs were issued for inherited metabolic disorders and 5 for Cystic Fibrosis. These figures are presented descriptively, without implying a direct correspondence between diagnoses identified by screening and RDPCs issued, as the card does not include age or date-of-birth variables that would allow unequivocal linkage. For some diseases in the PNRN panel, RDPCs were also issued in 2024 (in some cases covering all diagnoses in that year), whereas for Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD), only 1 card was issued out of 15 identified cases (**Table 2**).

The integration of neonatal screening with clinical registries is a well-established practice in some EU Member States. European reports document national programs with high coverage and centralized follow-up models post-screening, as observed in France[5] and Italy[6], which facilitate the transition to clinical registries as well as reference and planning activities.

[5] Bardou, M., et al. (2022). Newborn Screening Programmes: Review of the French Experience. *International Journal of Neonatal Screening*, 8(2). Available at: https://pmc.ncbi.nlm.nih.gov/articles/PMC9149820. [Accessed on 10 June 2025]

[6] Taruscio, D., et al. (2020). Expanded Newborn Screening in Italy: Update and Perspectives. *International Journal of Neonatal Screening*, 6(4). Available at: https://flore.unifi.it/handle/2158/1238376. [Accessed on 10 June 2025]

Table 2. Summary Overview – PNRN Diseases Corresponding to RDPCs in 2024

Rare Disease Included in PNRN	Confirmed Diagnoses (INSA 2024)	RDPCs Issued (RDPC 2024)
ORPHA232 - Sickle cell anemia	36	30
ORPHA586 - Cystic fibrosis	6	5
ORPHA42 – Medium chain acyl-CoA dehydrogenase deficiency	15	1
ORPHA33 – Isovaleric acidemia	1	1
ORPHA25 – Glutaryl-CoA dehydrogenase deficiency	1	1
ORPHA6 – 3-methylcrotonyl-CoA carboxylase deficiency	0	1
ORPHA943 – Malonic aciduria	0	1
ORPHA228302 – Carnitine palmitoyl transferase II deficiency, myopathic form	0	1

Methodological Note: *For this analysis, the list of rare diseases coded in RDPCs issued in the pediatric age group in 2024 was obtained, and it was verified which of these conditions are included in the screening panel of the National Neonatal Screening Program (in Portuguese, Programa Nacional de Rastreio Neonatal - PNRN). Subsequently, these diagnoses were cross-checked with confirmed positive cases reported by the PNRN in the same year (INSA 2024 data).*

The following tables (**Tables 3 and 4**) present the 10 rare diseases with the highest number of RDPCs issued in 2024 for the pediatric and adult age groups, respectively. They include two measures of relative frequency: (i) the proportion of the disease within the total RDPCs issued for the respective age group (1,500 cards for adults and 93 for pediatric age) and (ii) the proportion of the disease within the total coded diseases on the RDPCs issued for the age group (503 different diseases coded for adults and 40 for pediatric RDPCs).

Table 3. Top 10 rare diseases with the highest number of RDPCs issued in pediatric patients in 2024

TOP 10 – Rare Diseases in Pediatric Age	Number of RDPCs Issued	Relative Frequency within Total RDPCs Issued in Age Group (%)	Relative Frequency within Total Coded Diseases (%)
ORPHA232 - Sickle cell anemia	30	32,3%	75,0%
ORPHA636 - Neurofibromatosis type 1	7	7,5%	17,5%
ORPHA586 - Cystic fibrosis	5	5,4%	12,5%
ORPHA251365 - Sickle cell S-C disease	4	4,3%	10,0%
ORPHA98896 - Duchenne muscular dystrophy	4	4,3%	10,0%
ORPHA610 - Bethlem muscular dystrophy	3	3,2%	7,5%
ORPHA3205 - Sturge-Weber syndrome	3	3,2%	7,5%
ORPHA262 - Duchenne and Becker muscular dystrophy	3	3,2%	7,5%
ORPHA805 - Tuberous sclerosis complex	2	2,2%	5,0%
ORPHA166 - Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy	2	2,2%	5,0%

Table 4. Top 10 rare diseases with the highest number of RDPCs issued in adults in 2024

TOP 10 – Rare Diseases in Adults	Number of RDPCs Issued	Relative Frequency within Total RDPCs Issued in Age Group (%)	Relative Frequency within Total Coded Diseases (%)
ORPHA232 - Sickle cell anemia	107	7,1%	21,3%
ORPHA791 - Retinitis pigmentosa	66	4,4%	13,1%
ORPHA636 - Neurofibromatosis type 1	38	2,5%	7,6%
ORPHA169808 - Mild hemophilia A	28	1,9%	5,6%
ORPHA363700 - Neurofibromatosis type 1 due to NF1 mutation or intragenic deletion	25	1,7%	5,0%
ORPHA60 - Alpha-1-antitrypsin deficiency	25	1,7%	5,0%
ORPHA85447 - ATTRV30M amyloidosis	25	1,7%	5,0%
ORPHA589 - Myasthenia gravis	24	1,6%	4,8%
ORPHA166078 - Von Willebrand disease type 1	23	1,5%	4,6%

ORPHA327 - Congenital factor VII deficiency	22	1,5%	4,4%

In adulthood, sickle cell anemia emerged as the most frequent rare disease in the RDPC in 2024, with 107 cards issued, corresponding to 7.1% of the total RDPCs issued for adults. This hereditary hemoglobinopathy demonstrates high prevalence in specific regions of the world, namely Sub-Saharan Africa, the Middle East, India, and Brazil[7] [8]. The observed incidence in the RDPC may partly reflect recent migratory flows of adults from these regions, who access the National Health Service and, following diagnostic confirmation, obtain the respective card. This pattern correlates with European data linking the increase in adult diagnoses to international mobility, underscoring the need for public health policies adapted to demographic and epidemiological dynamics, as well as for adequate healthcare resources to address the growing prevalence of this condition[7] [9].

This trend justifies the continued epidemiological monitoring of rare diseases across different age groups, emphasizing the importance of integrating more granular demographic and clinical variables into the national RDPC database. Age-group segmentation will enable more robust longitudinal and comparative analyses, supporting the development of health policies targeted at both pediatric and adult populations. The consistent presence of diseases such as sickle cell anemia and neurofibromatosis type 1 across both age groups (**Tables 3 and 4**) suggests continuous clinical trajectories and enduring healthcare needs from childhood into adulthood. In the pediatric group, sickle cell anemia represents 32.3% of all RDPCs issued in this age group and 75% of all coded diseases, indicating a highly concentrated profile influenced by neonatal screening. In adults, diagnostic dispersion is greater, with no single disease exceeding 10% of the total RDPCs issued, demonstrating higher clinical heterogeneity.

Conversely, the concentration of specific diseases in only one age group highlights the importance of differentiated strategies for screening, diagnosis, and follow-up. The joint analysis of relative frequencies presented in each table allows a complementary interpretation of both the absolute burden of each disease within the total RDPCs issued for the group and its relative representation given the observed diagnostic diversity, providing a more comprehensive perspective on the epidemiological profile of each age group.

The convergence between national RDPC issuance data and prevalence data from Orphanet reinforces the utility of the RDPC as a complementary tool for epidemiological monitoring and health planning, considering its voluntary nature. It also demonstrates the positive impact of policies such as neonatal screening and the national network of Reference Centers on RDPC issuance, consolidating its role as a support tool for public health.

[7] World Health Organization (WHO). Sickle-cell disease: A strategy for the WHO African Region. Brazzaville: WHO; 2022. Available at: https://www.afro.who.int/publications/sickle-cell-disease-strategy-who-african-region. [Accessed on 10 July 2025].

[8] Orphanet. Sickle cell anemia – ORPHA:232. Available at: https://www.orpha.net/en/disease/detail/232. [Accessed on 10 July 2025]

[9] European Centre for Disease Prevention and Control (ECDC). Public health guidance on screening for sickle cell and thalassaemia in migrants. Stockholm: ECDC; 2018. Available at: https://www.ecdc.europa.eu/en/publications-data/public-health-guidance-screening-sickle-cell-and-thalassaemia-migrants. [Accessed on 10 July 2025]

Conclusion

In 2024, the Rare Disease Person's Card (RDPC) recorded a 12.4% increase in the number of cards issued compared with 2023, totalling 1,593 RDPCs and covering 515 distinct rare diseases, 117 of which were coded for the first time in Portugal. The cards were requested by 34 healthcare units across the country, with a significant concentration (82.7%) in seven Local Health Units (ULS) that host National Reference Centres, underscoring the strategic role of these institutions in the identification and clinical management of rare diseases. This growth reflects the commitment of healthcare professionals and the positive impact of targeted training initiatives, contributing to broader and more consistent use of the RDPC nationwide.

Despite the predominance of Reference Centres in RDPC issuance, the data reinforces the need to continue raising awareness among all medical specialties and hospital units regarding the existence and relevance of this card, given the cross-cutting nature of rare diseases and the unpredictability of emergency contexts in which timely access to clinical information may be critical.

Recently, the SNS Transparency Portal began publishing monthly updated information on RDPCs issued since 2020. This development enhances transparency and strengthens continuous monitoring capacity, enabling the analysis of temporal trends, geographical variations, and seasonality. It supports planning processes (resource allocation, training initiatives, communication strategies) and improves care delivery by increasing visibility of RDPC use within the national health system.

For 2025, several key targets have been defined, namely: enabling the visualization of the RDPC within hospital information systems at the point of triage in emergency contexts; automating the update of ORPHA codes based on Orphanet releases; integrating, whenever possible, demographic variables such as age and geographic location to allow more granular and informed analyses; promoting continuous professional training and participation in European initiatives, ensuring full compliance with the General Data Protection Regulation (GDPR) and the safeguarding of clinical information confidentiality; and maintaining the ORPHA nomenclature permanently up to date.

Achieving these objectives will reinforce the value of the RDPC as a tool supporting the delivery of personalized care, strengthening continuity of care, and contributing to a more effective, equitable, and person-centred response for individuals living with rare diseases.

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