RARE DISEASE PERSON'S CARD

Technical Report

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Table of Contents

List of Graphs, Tables and Figures

Resumo

O que é este documento?

O relatório anual sobre a implementação do Cartão da Pessoa com Doença Rara (CPDR), referente ao ano de 2025.

O que consta do documento?

Neste relatório anual são apresentados dados que demonstram a implementação do processo de requisição do CPDR no ano de 2025.

Quais são as principais conclusões?

Em cumprimento da Norma da DGS n.º 01/2018, verificou-se que durante o ano de 2025 foram emitidos 1803 CPDR, podendo observar-se o registo de 594 doenças raras diferentes nos novos cartões emitidos, das quais 155 são novas doenças codificadas pela primeira vez no ano de 2025. Trinta e quatro unidades de consultas de especialidade médica emitiram CPDR neste ano.

O que se quer atingir em 2026?

- 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 2025 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 2025.

What are the main conclusions?

In compliance with the DGS Guideline No. 01/2018, during 2024, 1803 RDPCs were issued, and 594 different rare diseases were registered, including 155 diseases registered for the first time in 2025. This year thirty-four specialized medical units have registered RDPCs.

What do we aim for 2026?

- 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;
- Expand and diversify the available indicators by incorporating regional and age-specific data, in order to enhance analysis and support evidence-based decision-making.

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 11798 RDPCs having been requested up to December 31, 2025.

Table 1. Rare Disease Person's Card Implementation

INDICATOR	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024	2025
Number of cards requested	622	911	776	2703	1100	1065	895	1142	1039	1417	1593	1803
Number of issuing healthcare units	6	13	14	24	30	25	24	25	30	35	34	34
Number of new diseases coded	168	182	106	225	150	145	90	97	82	110	117	155

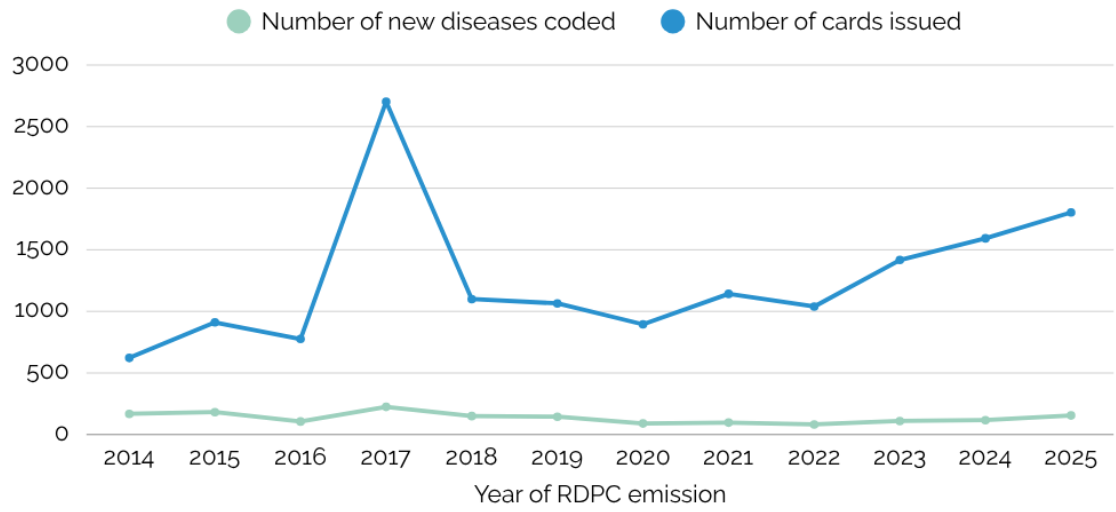
The number of new RDPCs issued in 2025 increased by 13,2% compared to 2024, which could be related to the training sessions conducted for physicians and facilitated by the Directorate-General of Health (DGS), within the scope of ORPHA codification and the European Project OD4RD (Orphanet Data For Rare 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.

However, comparing the number of new diseases coded with ORPHA codes in 2025 to the total of diseases coded in previous years, it is possible to conclude that 155 new rare diseases were coded for the first time in 2025 (**Graph 1**).

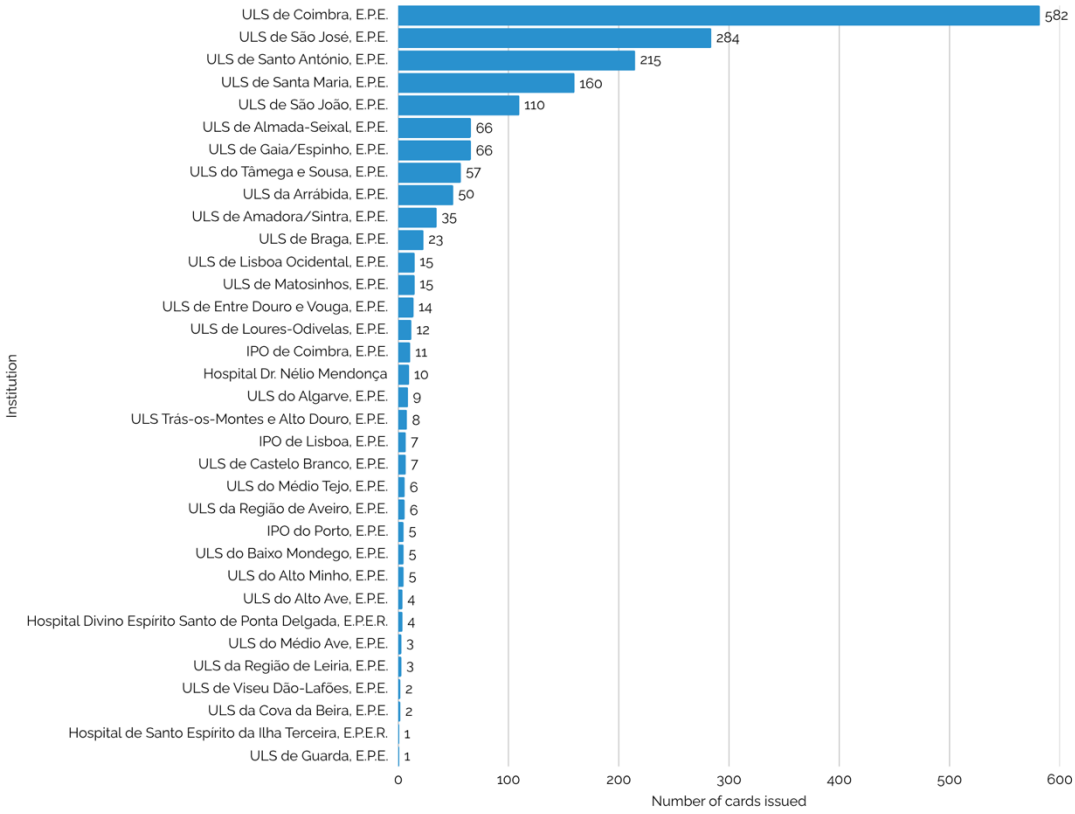
Graph 1. Evolution of the Implementation of Rare Disease Person's Card in Portugal



By analysing individual information by healthcare provider, it can be observed that, in 2025, the 1803 RDPCs requested were issued by 34 institutions, as shown in **Graph 2**, the same as in 2024. It was also verified that 77,3% of RDPCs were requested by seven Local Health Units (ULS) and by the three Portuguese Institutes of Oncology (IPO) – Porto, Lisbon and Coimbra - which include Reference Centres for rare diseases, namely:

a) *Unidade Local de Saúde de Coimbra, E.P.E. (32,3%);*
b) *Unidade Local de Saúde de São José, E.P.E. (15,8%);*
c) *Unidade Local de Saúde de Santo António, E.P.E. (11,9%);*
d) *Unidade Local de Saúde de Santa Maria, E.P.E. (8,9%);*
e) *Unidade Local de Saúde de São João, E.P.E. (6,1%);*
f) *Unidade Local de Saúde de Lisboa Ocidental, E.P.E. (0,8%);*
g) *Instituto Português de Oncologia de Coimbra Francisco Gentil, E.P.E. (0,6%);*
h) *Instituto Português de Oncologia de Lisboa Francisco Gentil, E.P.E. (0,4%);*
i) *Instituto Português de Oncologia do Porto Francisco Gentil, E.P.E. (0,3%);*
j) *Unidade Local de Saúde do Alto Ave, E.P.E. (0,2%).*

Graph 2. Number of RDPCs requested by Institution in 2025



Analysing the information by disease, it can be noted that in 2025 a total of 594 different rare diseases (594 distinct ORPHA codes) were recorded in the newly issued cards — corresponding to around 9-10% of the universe of rare diseases described in the Orphanet database. Of these, 155 represent new diseases coded for the first time in 2025.

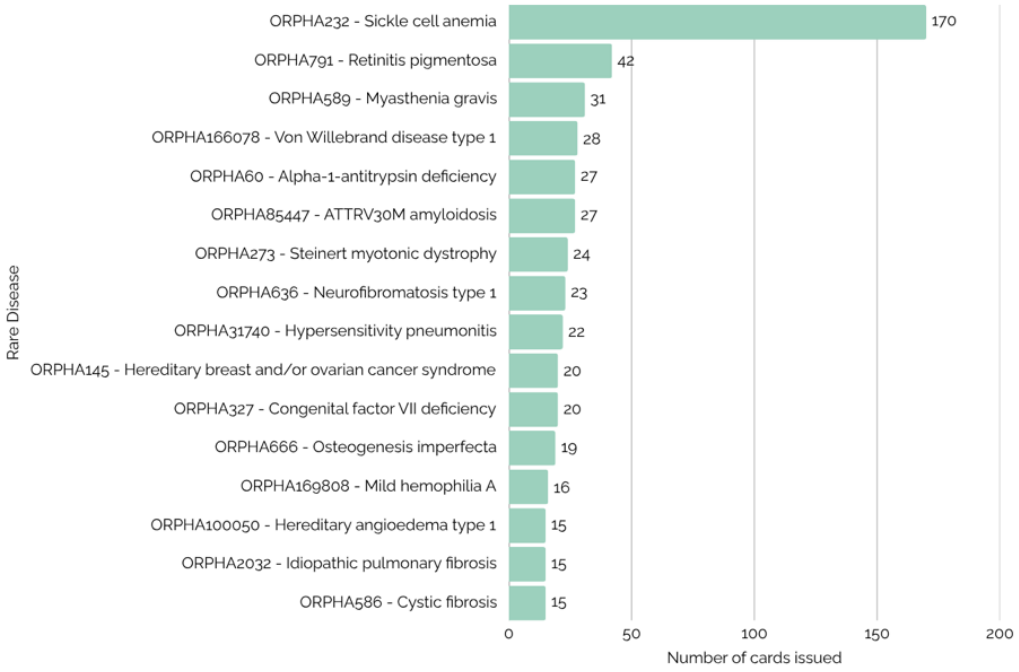
As in 2024, the rare diseases with the highest number of RDPC requests in 2025 were Sickle Cell Anemia and Retinitis Pigmentosa (**Graph 3**). These diseases are also among the most prevalent in Europe, according to the 2025 list published annually by Orphanet[2].

Following recent updates to the CPDR database, which included the large-scale removal of duplicate records and the consolidation of multiple diagnoses per person into a single card, historical values were revised, resulting in a reduction in the number of cards issued in previous years and, consequently, in the cumulative total. Between 2014, the year CPDR issuance began in Portugal, and the end of 2025, a total of 12784 CPDR cards were issued and 1627 different rare diseases were identified (**Table 1**).

[2] Orphanet Report Series (2025). 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 28/02/2026].

By the end of 2025, according to the analysis of the dataset[3] available on the Orphanet portal, 6102 ORPHA codes corresponding to rare diseases at the pathology classification level had been described, excluding codes referring to groups or subgroups of diseases.

Graph 3. Rare diseases with the most cards requested in 2025



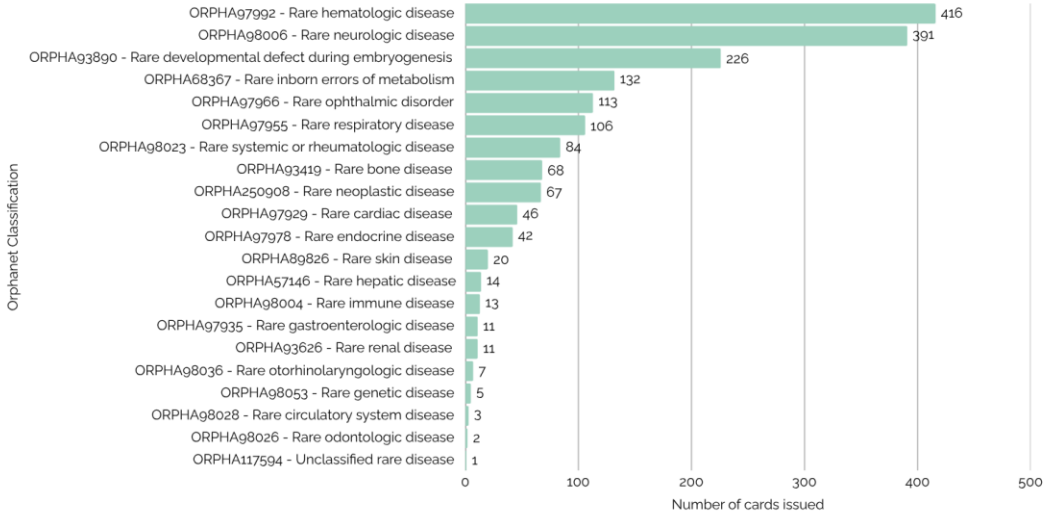
To provide a more detailed characterization of rare diseases recorded in RDPC issuance, cards issued in 2025 were grouped according to the Orphanet Classification, as shown in **Graph 4**. This categorization was based on the correspondence between the ORPHA codes of the cards and the corresponding nosological classification. It should be noted that, in this analysis, the total number of RDPCs by Orphanet classification (1845) exceeds the total number of cards actually issued (1803 in 2025). 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 2025, 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

[3] Orphanet (2025). Orphadata – *Natural History of Rare Diseases. Orphanet database of rare diseases*. Available at: https://sciences.orphadata.com/natural-history/ [Accessed on 8 March 2026].

rare ophthalmological diseases (e.g., Retinitis Pigmentosa). This epidemiological profile aligns with European prevalence data provided by Orphanet through Orphadata (December 2025 version)[4].

Graph 4. Number of RDPCs issued in 2025, by Group of Diseases according to the Orphanet Classification



[4] Orphanet Knowledge base release of December 2025. Epidemiology (prevalence and/or incidence) of Orphanet rare diseases. Available at: https://www.orphadata.com/epidemiology/ [Accessed on 28/02/2026]

Conclusion

In 2025, the Rare Disease Person's Card (RDPC) recorded a 13,2% increase in the number of cards issued compared with 2024, totalling 1803 RDPCs and covering 594 distinct rare diseases, 155 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 (76,6%) 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 2026, 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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