

Technical Report

RARE DISEASE PERSON'S CARD

2022

Department of Quality in Health



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Resumo

O que é este documento?

Relatório anual sobre a implementação do Cartão da Pessoa com Doença Rara (CPDR), referente ao ano de 2022, elaborado pelo Departamento da Qualidade na Saúde, Divisão de Planeamento e Melhoria da Qualidade.

O que consta do documento?

Neste relatório anual são apresentados dados que demonstram a implementação do processo de requisição do CPDR no ano de 2022.

Quais são as principais conclusões?

Em cumprimento da Norma da DGS n.º 01/2018, verificou-se que durante o ano de 2022 foram emitidos 1039 CPDR, podendo observar-se o registo de 387 doenças raras diferentes nos novos cartões emitidos, das quais 82 são novas doenças codificadas pela primeira vez no ano de 2022. Trinta unidades de consultas de especialidade médica emitiram CPDR neste ano.

O que se quer atingir em 2023?

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

Summary

What is this document?

The 2022 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 2022.

Main conclusions?

In compliance with the DGS Guideline No.01/2018, during 2022, 1,039 RDPCs were issued, and 387 different rare diseases were registered, including 82 diseases registered for the first time in 2022. This year thirty specialized medical units have registered RDPCs.

What do we aim for 2023?

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

Introduction

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

Rare diseases exhibit the following characteristics:

- a) In general, 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, but the appropriate treatment and medical care can improve the quality of life of those affected and extend their life expectancy;
- 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.

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 2013, 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.

This report presents the progress of the RDPC implementation process, with an emphasis on monitoring data for the year 2022.

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 a European 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 30/06/2020].

The RDPC should be requested by a physician through the electronic patient record on the Portuguese Health Data Sharing Platform (Plataforma do Registo de Saúde Eletrónico), which provides a list of rare diseases with their correspondent ORPHA codes and clinical guidelines in emergent or urgent situations. The physician can edit the clinical guidelines and, if necessary, personalize and adjust the information according to each individual patient's case, enabling the customization of urgent/emergency care.

In order to simplify the process and clarify the RDPC issuance procedure, the DGS guideline ([DGS Norma No. 01/2018](#)) defines its emission and review conditions.

Results of the RDPC Implementation Process

Over the last 9 years, as observed in Table 1, healthcare professionals and units have shown increasing interest in requesting RDPCs, with 10,253 RDPCs having been requested until 31 December 2022.

Table 1. Rare Disease Person's Card Implementation.

Indicator	2014	2015	2016	2017	2018	2019	2020	2021	2022
No. of Cards Requested	622	911	776	2703	1100	1065	895	1142	1039
No. Health Care Units Issuing	6	13	14	24	30	25	24	25	30
No. New Coded Diseases	168	182	106	225	150	145	90	97	82

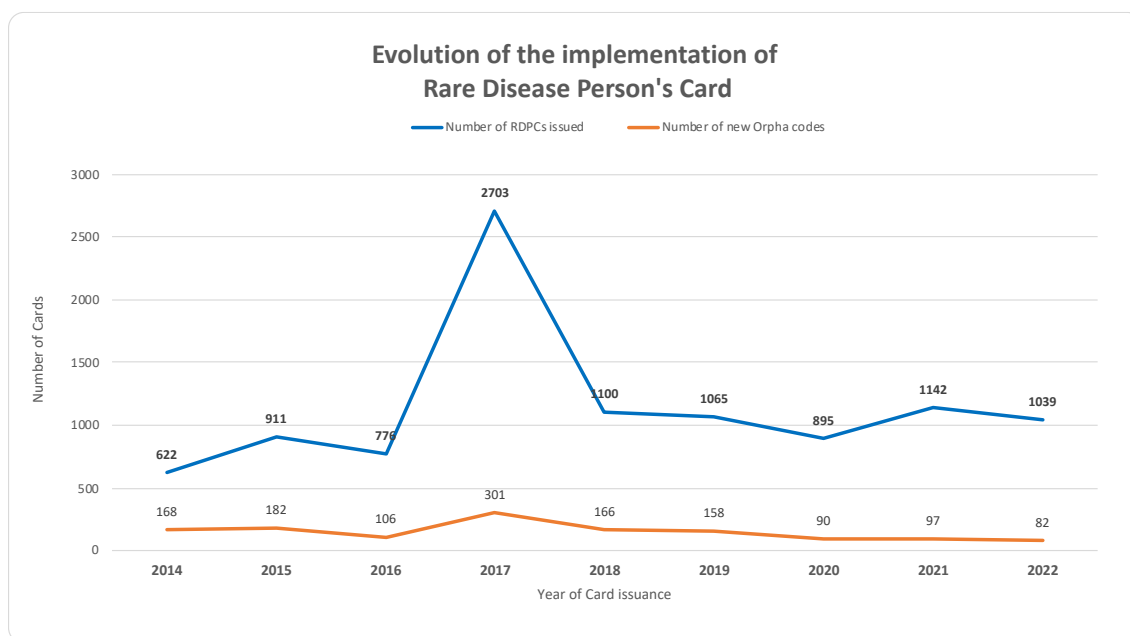
Source: DGS, based on data provided by SPMS in 2023. The number of cards requested each year has been revised and updated.

The number of new RDPCs issued in 2022 slightly decreased compared to 2021. In contrast, 2017 data indicate a significant increase in the number of cards requested, highly related to the expanded possibility of requesting RDPC to all public and private hospitals, that are 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:

- Obtaining a rare disease diagnosis is mostly a complex and lengthy process, the reason why diagnoses are usually only made by Reference Centres.
- 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.
- Recently constraints in updating the available ORPHA codes in RDPC.

However, comparing the number of new diseases coded with ORPHA codes in 2022 to the total of diseases coded in previous years, it is possible to conclude that 82 new rare diseases were coded for the first time in 2022.

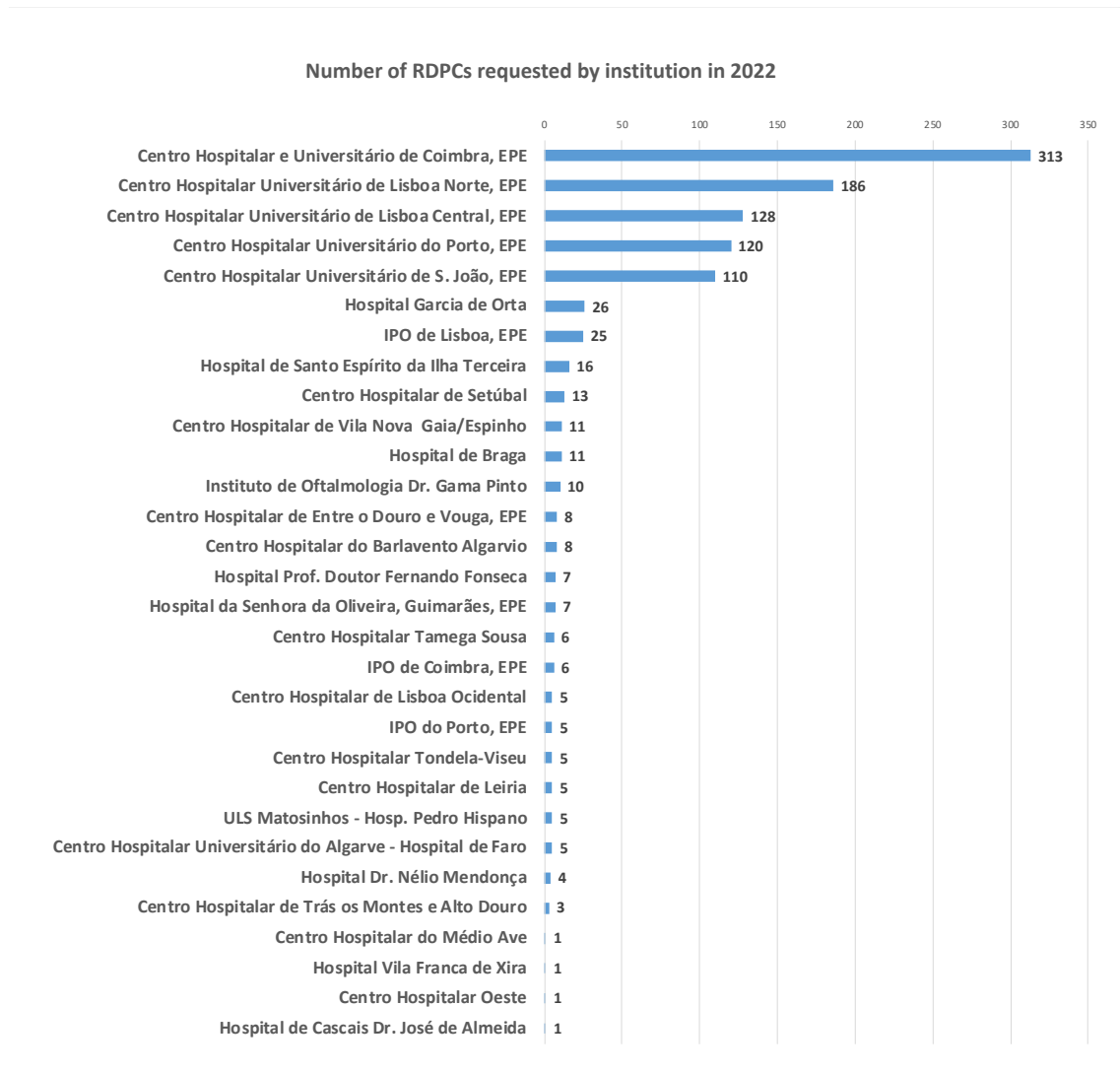
Chart 1. Evolution of the Implementation of Rare Disease Person’s Card.



Source: DGS, based on data provided by SPMS in 2023.

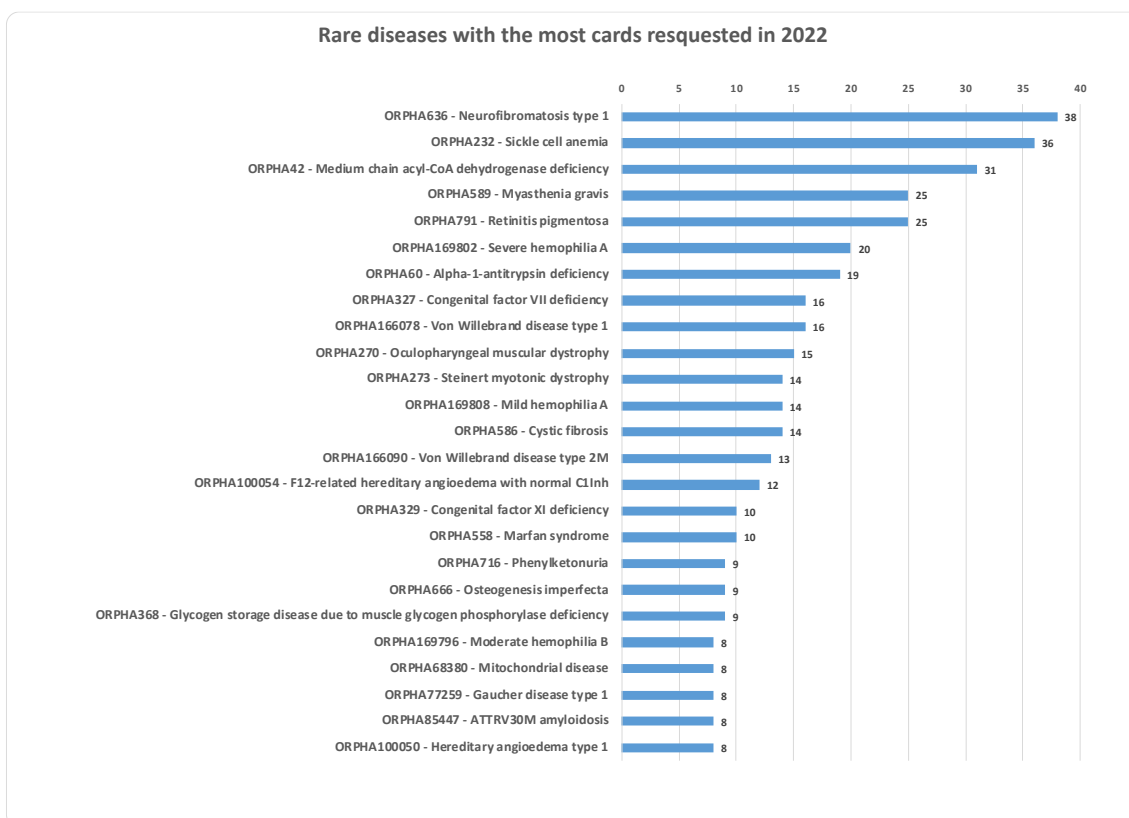
By analysing individual information by the healthcare provider (Chart 2), it can be observed that, in 2022, the 1,039 RDPCs requested were issued by 30 institutions, five more institutions than in the previous year. Also, 85.0% of RDPCs were requested by eight institutions with Reference Centres for rare diseases, namely *Centro Hospitalar Universitário de Coimbra* (29.8%), *Centro Hospitalar Universitário de Lisboa Norte* (17.7%), *Centro Hospitalar Universitário de Lisboa Central* (12.2%), *Centro Hospitalar Universitário do Porto* (11.4%), *Centro Hospitalar Universitário de S. João* (10.5%), *Hospital da Senhora da Oliveira – Guimarães* (0.7%), *IPO de Lisboa* (2.4%), and *IPO do Porto* (0.5%).

Chart 2. Number of RDPs requested by each institution in 2022.



Source: DGS, based on data provided by SPMS in 2023.

Chart 3. Rare Diseases with the highest number of RDPCs requested in 2022.



Source: DGS, based on data provided by SPMS in 2023.

Analysing the information by disease, it can be noted that 387 different rare diseases (387 distinct ORPHA codes) were recorded on the new cards issued, including 82 diseases registered for the first time in 2022.

In 2022, the 1,039 cards issued corresponded to 1,035 persons living with a rare disease. Since RDPC has been established, in early 2014, until the end of 2022, 1,245 different rare diseases have been identified (cf. Table 1), corresponding to 8,865 persons with rare diseases holding RDPCs.

Over time, the issuance of a new card for the same patient may occasionally occur. On one hand, this fact can be associated with updating the preliminary diagnosis to a more accurate one, corresponding to an updated designation of the rare disease, different from the initial one. On the other hand, it may be related to the identification of an additional and distinct rare disease.

The rare diseases with the highest number of RDPCs requested in 2022 were Neurofibromatosis type 1, Sickle Cell Anemia, Medium Chain Acyl-CoA Dehydrogenase Deficiency, Myasthenia Gravis, Severe Hemophilia A, and Retinitis Pigmentosa (Chart 3).

Conclusion

During 2022, 1,039 RDPCs were issued, and 387 different rare diseases were registered, including 82 diseases registered for the first time. These RDPCs were requested from different 30 health units across the country, with 85.0% being requested by eight institutions with Reference Centres for rare diseases, namely *Centro Hospitalar Universitário de Coimbra* (29.8%), *Centro Hospitalar Universitário de Lisboa Norte* (17.7%), *Centro Hospitalar Universitário de Lisboa Central* (12.2%), *Centro Hospitalar Universitário do Porto* (11.4%), *Centro Hospitalar Universitário de S. João* (10.5%), *Hospital da Senhora da Oliveira – Guimarães* (0.7%), *IPO de Lisboa* (2.4%), and *IPO do Porto* (0.5%).

Although hospital units with Reference Centres for rare diseases are the institutions that issue the highest number of RDPCs, there is a need to continue promoting the existence of this tool in all specialty consultations since rare diseases can occur in all different age groups throughout the country and the access of tailored care can arise at any emergency department.

For 2023, to enhance the usefulness and dissemination of RDPC, DGS maintains the following proposals to be implemented by the Shared Services of the Ministry of Health (SPMS – Serviços Partilhados do Ministério da Saúde, EPE):

- Publication of ORPHA Coding in the National Clinical Terminologies Centre;
- Facilitate the display of RDPC in the information systems of Hospital Emergency Departments during the triage process, to ensure healthcare professionals have prompt access to relevant information regarding people living with a rare disease, the specifics of their clinical conditions, and the care needed, in emergency situations.
- Automatic integration of ORPHA code updates into RDPC, provided by the Orphanet National Team.



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