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Introduction

Since the beginning of the Coronavirus Disease of 2019 (COVID-19) pandemic, countless difficulties have been affecting the daily living of people worldwide. Strict measures of social distancing, personal protective equipment mandates, restricted access to healthcare, impact on economic stability and mental health are all concerns that still affect many people.¹

People living with rare diseases, like RASopathies, are a particularly vulnerable community greatly impacted by the pandemic. The burden of these challenges can be exacerbated in a country like Colombia with growing economic disparities, an unequitable health system, and delayed access to SARS-CoV-2 vaccination.² Our aim was to assess the experiences of RASopathies patients and their families on how the COVID-10 pandemic challenges have impacted their lives, in order to increase public and scientific awareness on the topic.

Methods

Observational cross-sectional study with a convenience sampling strategy between April 1st and May 15th, 2021. A questionnaire adapted from previous published research on COVID-19 pandemic and quality of life or related publications was designed. It was addressed to patients/caregivers living with a RASopathy, who were contacted via phone call. Twenty replies were obtained. None of the replies were excluded. Descriptive statistical analysis was performed. Absolute frequencies and percentages were used.

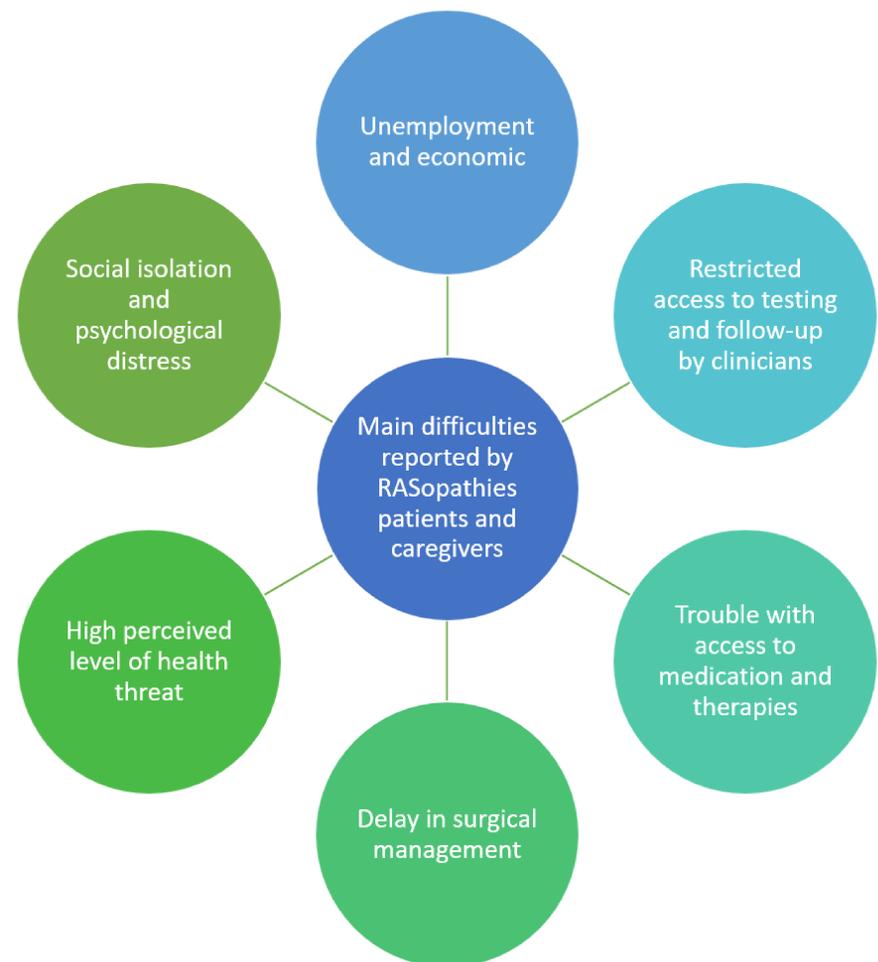
Results

A total of 20 individuals (19 caregivers and 1 patient) were included in this study. All of them live in the south-west region of Colombia, with the exception of two individuals from the Amazonas department (far south of Colombia). The majority of patients (14/20, 70%) have a diagnosis of Noonan syndrome (NS), followed by neurofibromatosis type 1 (4/20, 20%), NS with multiple lentiginos (1/20, 5%) and NS with loose anagen hair (1/20, 5%).

Household composition was primarily of three or four individuals, with one patient diagnosed with a RASopathy (17/20, 85%). Employment was distributed as: employed (11/20, 55%), stay-at-home parent (5/20, 25%), unemployed and fit to work (3/20, 15%), and other status (2/20, 10%). Restricted access to testing and follow-up by specialist was reported by the majority of individuals (15/20, 75%).

Difficulty to schedule an appointment with a geneticist was widely reported (11/20, 55%), and access to telemedicine greatly varied between specialties, with most patients having a positive experience (7/9, 77.7%).

Figure 1. RASopathies patients and caregivers reported difficulties associated with the COVID-19 pandemic



Trouble with access to medication for those with pharmacologic treatment was stated (5/16, 31.2%), and with access to therapies (12/16, 75%). Delay in surgical management was common (7/10, 70%).

Most caregivers reported leaving their home only for essential activities (12/20, 60%), while the rest stated to leave the house daily for work. Almost all caregivers described the level of health threat as very high or high for the patients and reported full adherence to mask mandate (18/20, 90%). Generally, families felt that economic difficulties, social isolation, and restricted healthcare access were the main challenges for patients during the pandemic (19/20, 95%). 5 caregivers have been able to receive a vaccine against SARS-CoV-2 (26.3%). Only 2 individuals reported hesitancy to vaccination.

Conclusions

RASopathies patients and caregivers have been greatly affected by the economic difficulties, social isolation, and restricted healthcare access that the COVID-19 pandemic has caused.

Health disparities and limited access to vaccination can delay solutions to these challenges.³ Important measures need to be implemented in order to address these limitations and provide patients with the multidisciplinary care they need.

References

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