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COVID-19 and the rare disease organization response during pandemic: the ‘Italian model’

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“The ability of a community to ‘transform a resilience pattern in good practice’ is a key factor, helping the healthcare system to be ready for future catastrophic events.”

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On 31 January 2020, the Italian government declared a state of emergency due to SARS-CoV-2 and in March 2020 placed more than 60 million people in lockdown. At the peak of the pandemic, Italy’s number of active cases was one of the highest in the world [1]. Hospitals were overwhelmed by COVID-19 patients, or even forced to close because of diminishing staff. Consequently, obtaining treatment (including acute care and outpatients access) was compromised for thousands of patients affected by rare diseases (RDs). This scenario highlighted the substantial healthcare burdens and social issues that are typical of RDs, even beyond the pandemic emergency: poor understanding of all RDs (exceeding 7000 conditions) requiring specialized physicians; early access to specific treatments (when available), practicable healthcare policies and avenues of management from referral centers to the territorial hospitals and outpatients services. As much as 10% of all hospital admissions in Western Australia are related to RDs and cost (i.e., RDs accounted for 10.5% of total hospital expenditure: US\$395 million per year) [2].

An online survey was launched (23 March–5 April 2020) by the National Centre for Rare Diseases of the Istituto Superiore di Sanità (ISS) and UNIAMO. The survey was circulated through the Telefono Verde Malattie Rare helpline of the ISS and diffused via social media such as Facebook, LinkedIn, Twitter and Instagram. There were 31 items regarding the healthcare issues encountered during the first wave of the pandemic. All the Italian regions have participated, with the higher rate of response in Lombardy, Lazio and Tuscany. The survey was active for a 2-week period, from 23 March to 5 April 2020, and a total of 1174 surveys was collected. Patients and caregivers were invited to express their medical and special needs during the pandemic. The main difficulties concerned both healthcare services and social support: 52% of the patients avoided any medical inpatient or outpatient consult, or quit physical rehabilitation and physiotherapy; 16% encountered drug access difficulties [3]. The responses also highlighted a significant need for information regarding the pandemic’s impact on the health condition of RD patients [3–5].

What was the reaction of the Italian RD community at large? The Italian RD network, including the National Centre for Rare Diseases-ISS, the regional coordinator centers, together with UNIAMO and all patient associations developed resilience efforts, when different stakeholders, including politics, economic and health system and the communities, share the same priorities and work together. For instance, the NRCD-ISS and UNIAMO organized weekly webinars with national and international experts and patient associations to share information and good practices, including telemedicine and home therapy. Following the experience of the European Reference Networks for RDs, clinicians from major hospitals and regional centers were already acquainted with the teleconsulting systems [6] during the pandemic. ISS guidelines were promptly established to help clinicians using appropriately these new tools [7]. Home therapy and delivery were approved nationally, while regional centers and hospitals followed this protocol, on a case by case basis.

What's next? The ability of a community to 'transform a resilience pattern in good practice' is a key factor, helping the healthcare system to be ready for future catastrophic events. With the next 'National Rare Disease Plan' and a dedicated RD nations law in progress, a number of key steps should be prioritized: 'information needs to travel, not the patient': telemedicine programs need to be developed in the chronic and acute settings (emergency), using e-monitoring systems (e.g., respiratory system – e-spirometers; heart-ECG; glycemia monitoring; telete habilitation etc.) to reach the patient home and to avoid unnecessary hospital admissions; the connection between the hub hospital systems, the district and community hospital (close to the patient's territory) needs to be reinforced: in the Piano Nazionale Ripresa e Resilienza (PNRR) e Piano Nazionale Prevenzione (PNP), the development of specific avenues for chronic and end stage diseases, the development of multidisciplinary care units with specific case managers (i.e., general practitioners and specialized nurses) and the development of specific 'assistance and care community houses' ('case della comunità'), represent an important novelty in the national and regional care models ('home care model': reaching the patients with the individual needs (i.e., infusion therapies, nutrition, ventilation, life-support, etc); early diagnosis means early management: the new technology developments, such as big data use, machine learning and artificial intelligence algorithms, will help medical doctors assess clinical pathways and transform suspects in diagnosis and management [8,9].

Author contributions

G Limongelli, D Taruscio and A Scopinaro contributed to the conception and design of the work. G Limongelli and D Taruscio drafted the manuscript. All the authors critically revised the manuscript, gave final approval and agree to be accountable for all aspects of work ensuring integrity and accuracy.

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References

1. Our World In Data. Daily new confirmed COVID-19 deaths per million people. World, Italy. <https://ourworldindata.org/explorers/coronavirus-data-explorer?zoomToSelection=true&pickerSort=desc&pickerMetric=location&Metric=Confirmed+deaths&Interval=7day+rolling+average&Relative+to+Population=true&Align+outbreaks=false&country=EuropeanUnion~ITA~OWID.WRL>
2. Garcia M, Downs J, Russell A *et al.* Impact of biobanks on research outcomes in rare diseases: a systematic review. *Orphanet J. Rare Dis.* 13, 202 (2018).
3. Taruscio D, Bertinato L, Carta C *et al.* Census of needs (March 23–April 5, 2020) of people with rare diseases in the current SARS-CoV-2 emergency scenario. <https://www.iss.it/documents/20126/0/Rapporto+ISS+COVID-19+n.+39+EN.pdf/0e65f2d5-5c93-d737-84af-e9dd0edc64e2?t=1620987775521>
4. Limongelli G, Iucolano S, Monda E *et al.* Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Registry. *J. Public Health (Oxf)*. fclab137 (2021). online ahead of print
5. Monda E, Iucolano S, Galdo M *et al.* Prevalence and direct costs of patients with neuromyelitis optica: data from Campania region in southern Italy. *Future Rare Dis.* 1(4), FRD17 (2021).

6. Mönig I, Steenvoorden D, de Graaf JP *et al.* CPMS-improving patient care in Europe via virtual case discussions. *Endocrine* 71(3), 549–554 (2021).
7. Gabbrielli F, Bertinato L, De Filippis G *et al.* Interim provisions on telemedicine healthcare services during COVID-19 health emergency. Istituto Superiore di Sanità, Roma, Italy (2020). <https://www.iss.it/documents/20126/0/Rapporto+ISS+COVID-19+n.+12+EN.pdf/14756ac0-5160-a3d8-b832-8551646ac8c7?t=1591951830300>
8. Hirsch MC, Ronicke S, Krusche M *et al.* Rare diseases 2030: how augmented AI will support diagnosis and treatment of rare diseases in the future. *Ann. Rheum. Dis.* 79, 740–743 (2020).
9. Benfredj R. How healthtech is transforming the future of rare disease diagnosis. *Future Rare Dis.* 1(1), FRD3 (2021).

