The DM-Scope registry: an innovative framework for myotonic dystrophy translational research

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BACKGROUND

The relevance of registries as a key component for developing clinical research for rare diseases (RD) and improving patient care has been acknowledged by most stakeholders. As recent studies pointed to several limitations of RD registries our challenge was (1) to improve standardization and data comparability; (2) to facilitate interoperability between existing RD registries; (3) to limit the amount of incomplete data; (4) to improve data quality. DM-Scope registry was developed to achieve these objectives for Myotonic Dystrophy (DM), a prototypical example of highly heterogeneous RD. Over the last few years we have contributed to several TGDOC meetings to define the mode of interaction between DM-Scope Registry and TREAT-NMD.

METHODS

The DM-Scope registry was developed in France in 2008. The main objective was to increase the epidemiological knowledge in DM, to harmonize patients medical follow-up, and to facilitate selection and enrolment of DM patients in clinical trials, particularly in a multicentre setting.

DM-Scope registry collects relevant clinical and epidemiological data on a standardized form during routine medical evaluation performed in French neuromuscular reference centres. Only patients with confirmed genetic diagnosis were included and data was collected by health-care practitioners. The DM-Scope form is shared with the Quebec registry to promote international research.

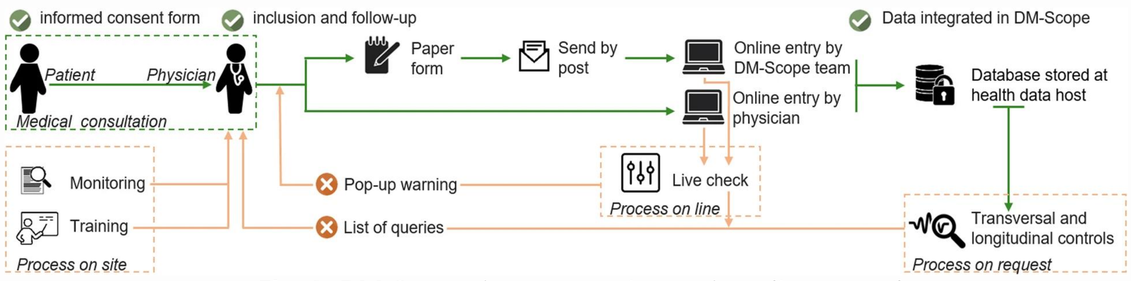

Fig. 1. DM-Scope data processing and quality control.

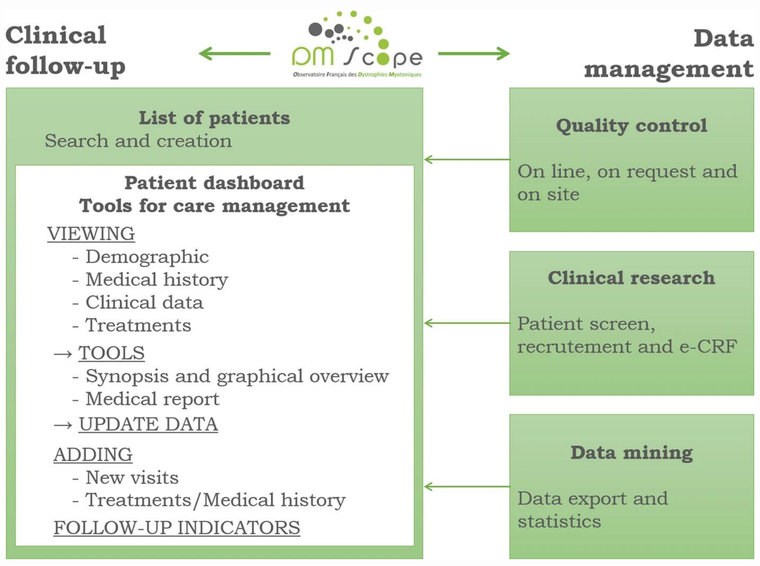

Fig. 2. Functionalities and database interface.

RESULTS

The dataset includes social-demographic data, clinical features, genotype, and biomaterial data, and is adjustable for clinical trial data collection. The registry has a nationwide coverage, composed of 55 neuromuscular centres and gathers almost 3000 DM patients (DM1 *n* = 2828, DM2 *n* = 142), both children (*n* = 322) and adults (*n* = 2648), which accounts for > 20% of overall registered DM patients internationally.

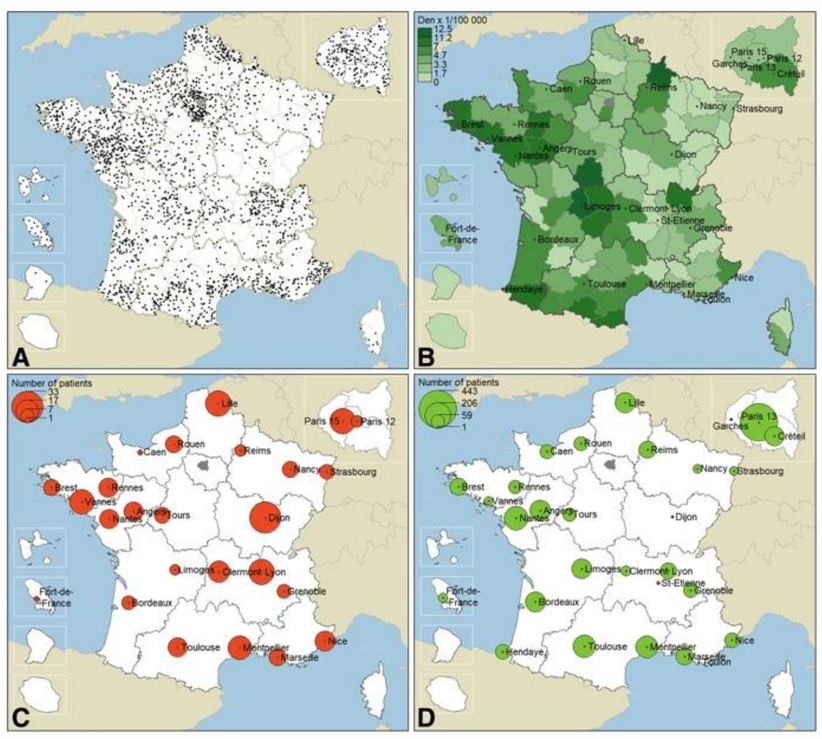

Fig. 3. Cartography of place of residence of enrolled DM participants. **A** - The individual representation (*N* = 2875). **B** - The regional distribution according to the density of population (N = 2875). **C** - Distribution of DM-Scope enrolled patients among paediatric French neuromuscular expert centres (26 centres, *N* = 255). **D -** Distribution of DM-Scope enrolled patients among adult French neuromuscular expert centres (29 centres, *N* = 2620).

The DM-scope registry has enrolled almost 3000 DM patients since 2008 (Fig. 4). Inclusion of the 2970 patients has been regular up to now (green line). The collected data have been annually updated since 2010. Between 2008 and 2018, more than half of the enrolled DM patients (53.3%) have been followed-up at least once, 30.9% at least twice, and 17.9% at least three times.

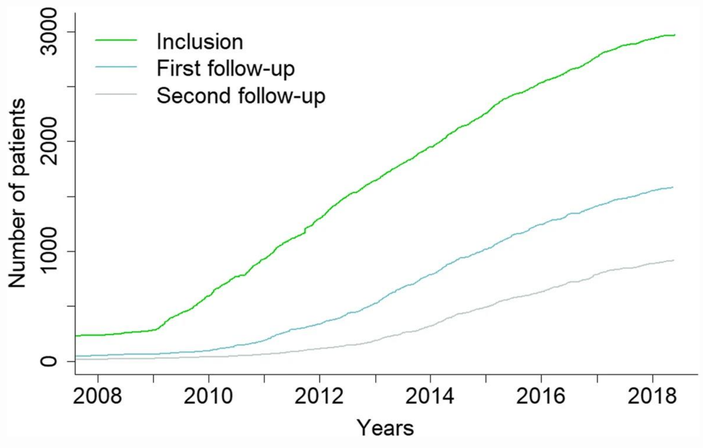

Fig. 4. Cumulative number of participants in the DM-Scope Registry.

CONCLUSION

The DM-Scope registry represents the largest collection of standardized data for the DM population. Our concept improved collaboration among health care professionals by providing annual follow-up of quality longitudinal data collection. The combination of clinical features and biomolecular materials provides a comprehensive view of the disease in a given population. The registry proves to be a powerful device for promoting both research and medical care that is suitable to other countries. In the context of emerging therapies, such integrated platform contributes to the standardisation of international DM research and for the design of multicentre clinical trials. Finally, this valuable model is applicable to other RDs.

Website : **www.dmscope.fr**.