An Overview of the CureDRPLA Global Patient Registry – Collecting Patient Reported Data to Advance Research

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BACKGROUND

- Dentatorubral-pallidoluysian atrophy (DRPLA) is a very rare ataxia.
- Inherited in an autosomal dominant manner and caused by expanded CAG repeats in the atrophin-1 gene[1].
- DRPLA is a clinically heterogeneous disease where clinical manifestations differ according to age at onset and CAG repeat size [1-3].
- Juvenile-onset cases (< 20 years old, with ≥ 65 repeats) typically present with the progressive myoclonus epilepsy phenotype [1-3].
- Adult-onset cases (≥ 20 years old, with < 65 repeats) present with variable combinations of ataxia, choreoathetosis, dementia and psychiatric symptoms [1, 3].
- The patient advocacy groups CureDRPLA and Ataxia UK have established the CureDRPLA Global Patient Registry to collect patient-reported data on individuals affected with DRPLA.

OBJECTIVES

The CureDRPLA Global Patient Registry will:

- Characterise and describe the DRPLA population.
- Enhance the understanding of prevalence across the world.
- Provide patient-reported data and burden of illness to researchers.
- Document differing patterns of diagnostic journeys for DRPLA patients.
- Connect participants with opportunities to take part in research.

METHODS

- Online registry available in 6 languages to facilitate global participation (English, Japanese, Korean, Portuguese, French, and Italian).
- Completed by individuals with DRPLA, caregivers and/or family members.
- Upon enrolment, participants are asked to consent and answer questions about demographics, diagnosis, medical history, activities of daily living, mobility, research, and disease and economic burden.
- Participants are asked to complete the registry once a year.
- Ethics committee approval was obtained (WCG IRB).

RESULTS

Overview of all participants

A **35** participants in total

B

Sex (M:F)	17:18
Age (mean ± SD)	30 ± 18.56
Age (min, max)	5, 68

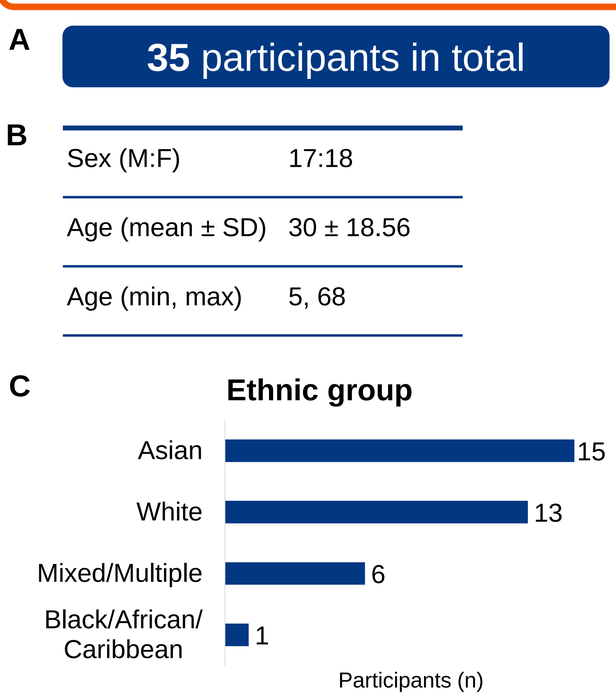

Figure 1. Overview of all the registry participants. A) Total number of participants in the CureDRPLA Global Patient Registry (n=35). B) Ratio of male and female participants and age distribution. C) Number of participants per ethnic group. D) Number of participants per country. E) Distribution of symptomatic and pre-symptomatic participants.

Juvenile- vs. adult-onset cohorts

		Juvenile- onset (< 20 years)	Adult-onset (≥ 20 years)
A	**Participants (n)**	20	11
B	**Age at symptom onset (mean ± SD)**	7 ± 4.52	39 ± 15.77
C	**# CAG repeats (mean ± SD)**	67 ± 7.71 (n = 17)	59 ± 2.07 (n = 5)
D	**Most common symptom attributed to disease onset (%)**	• 40% balance problems/ trouble walking or running • 40% epileptic seizures	• 73% balance problems/ trouble walking or running
E	**Present health concerns (i.e. last month) (%)**	• 85% balance problems/ trouble walking or running • 75% coordination problems in hands/arms and manual dexterity • 70% epileptic seizures • 70% intellectual disability	• 73% balance problems/ trouble walking or running • 64% coordination problems in hands/arms and manual dexterity • 46% mood swings/anxiety • 46% personality changes
F	**Experience epileptic seizures (%)**	80% present epilepsy	15% present epilepsy
G	**Strategies identified as helpful to manage DRPLA or DRPLA symptoms (%)**	• 40% medications or supplements • 35% physiotherapy • 30% occupational therapy	• 45% exercise (cardio or strength training) • 36% none • 27% medications or supplements
H	**Assistive care provided by**	• 100% parent • 47% professional caregiver • 37% sibling	• 50% spouse • 30% no assistance needed • 20% other relative

Table 1. Characterisation of the juvenile- and adult-onset cohorts. Responses analysed based on the age of symptom onset (Juvenile-onset < 20 years and adult-onset ≥ 20 years). A) Total number of participants per cohort. B) Average response to "At what age did you/the patient experience the first noticeable symptoms for DRPLA?" – number field. C) Average response to "What number of CAG repeats are present in the ATN1 gene?" – number field. D) Responses to "What were the first symptom(s) you/the patient experienced, even prior to the DRPLA diagnosis?" – multiple choice. E) Responses to "Which of the following DRPLA-related health concerns do you/the patient currently have (i.e. last month)?" – multiple choice. F) Responses to "Have you/the patient ever experienced epileptic seizures?" - Yes/no answer. G) Responses to "What have you/the patient tried to do to help manage DRPLA or DRPLA symptoms that you/the patient found was helpful?" – multiple choice. H) Responses to "Who provides assistive care for you/the patient when needed?" – multiple choice.

DISSEMINATION ACTIVITIES

We have publicised this registry broadly:

- 60 organisations for medical professionals (e.g. AAN, ABN).
- 91 patient organisations (e.g. NAF, Euroataxia, Epilepsy Foundation of America).
- 207 authors that published about DRPLA in the last 20 years.
- +1,000 medical professionals.
- Dissemination activities reached 86 countries.

CONCLUSION

- This registry is creating a cohort of well-characterised DRPLA patients for participation in future research studies.
- In time, it will also enhance the understanding of DRPLA prevalence across the world.

Thank you to all the individuals with DRPLA and families that participated!

SCAN ME

Help us share this registry!

Contact us

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References

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