

Introducing JOIN-HD: The Juvenile Onset Initiative for HD

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Background

Huntington's Disease (HD) is a rare inherited neurodegenerative disorder with a typical onset between the ages of 30 – 50. Juvenile onset Huntington's Disease (JoHD), defined by onset of symptoms before the age of 21, manifests differently from adult-onset HD. JoHD is present in approximately 5% of HD cases, although the exact prevalence is unknown.

Extensive research into the clinical and biological manifestations of JoHD have not occurred. Due to this lack of knowledge and awareness, JoHD presents unique challenges not only for the individuals affected but also for caregivers, family members, and healthcare professionals.

Design

JOIN-HD is a prospective, observational, multinational registry of individuals affected by JoHD. The primary objective is to identify and map the locations of affected individuals. Secondary objectives include supporting focused research for this population and identifying unmet needs to improve advocacy, care and support.

Participants will be referred for participation from HDYO's service user network, HD charities and family associations, professional networks and specialty clinics/physicians. Participants may also hear about the study through websites, social media, advocacy newsletters and support groups, or through snow-ball sampling. JOIN-HD will be carried out in three stages, as shown in Figure 1.

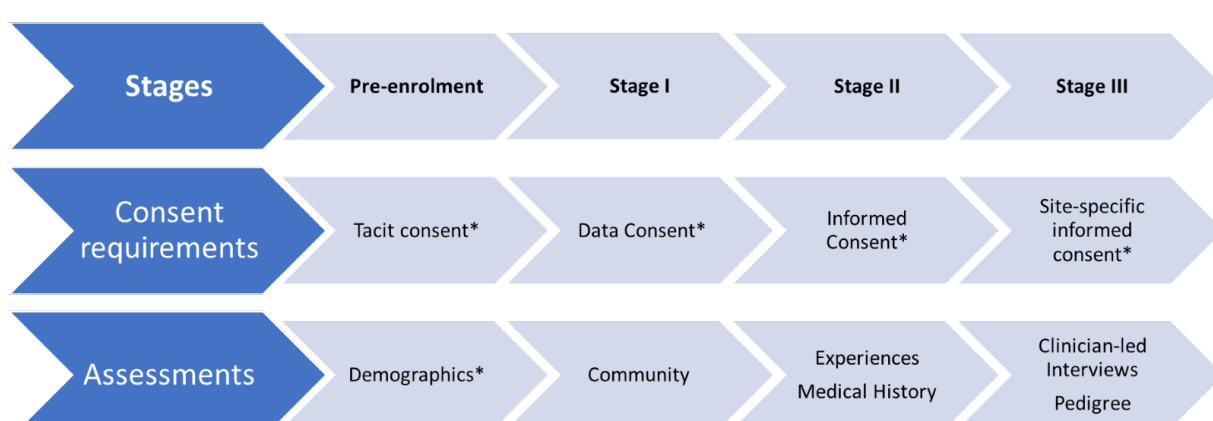


Figure 1: JOIN-HD stage diagram

Pre-registration

Pre-registration opened in March 2021, with potential participants registering their interest using Microsoft Forms. Currently, there are 17 pre-registrations from 6 countries, as shown in Figure 2.

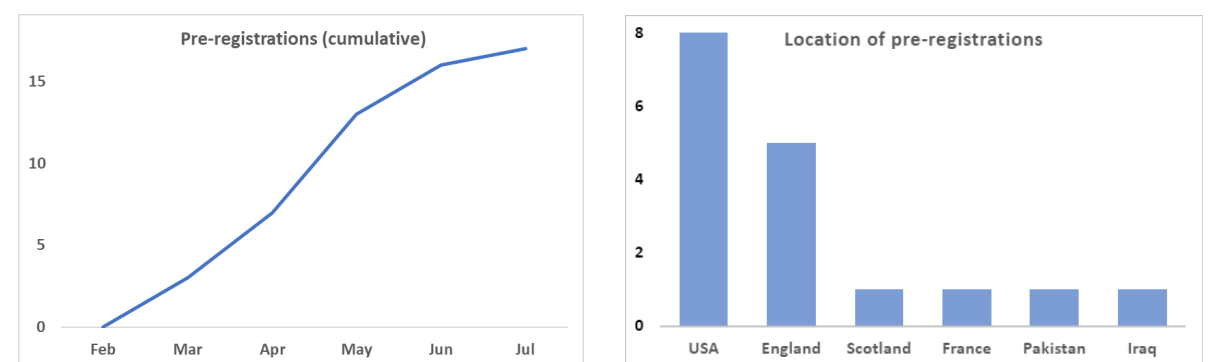


Figure 2: Number and location of JOIN-HD pre-registrations

Future stages

Stage I of JOIN-HD is due to launch later this year. Pre-registered participants will have a screening call with the JOIN-HD coordinator before being invited to self-enrol and participate remotely via an electronic data capture portal.

Stage I will capture demographic data and information about the links participants have with the HD community. At Stage II, following an informed consent process, data will be collected on participants' experience of JoHD. Stage III will incorporate a Clinician-led interview, where information on JoHD clinical characteristics, medical history, CAG repeats, current therapies and family history will be collected.

Conclusion

There is a clear need for increased research into JoHD, highlighted by the removal of the European Medicines Agency paediatric class waiver for HD in 2015. The JOIN-HD registry aims to increase understanding of JoHD and facilitate future research through the identification of potentially eligible participants.