Empowering Patients in Research: First insights from 444 Participants in the PATRE SYNGAP1 Registry

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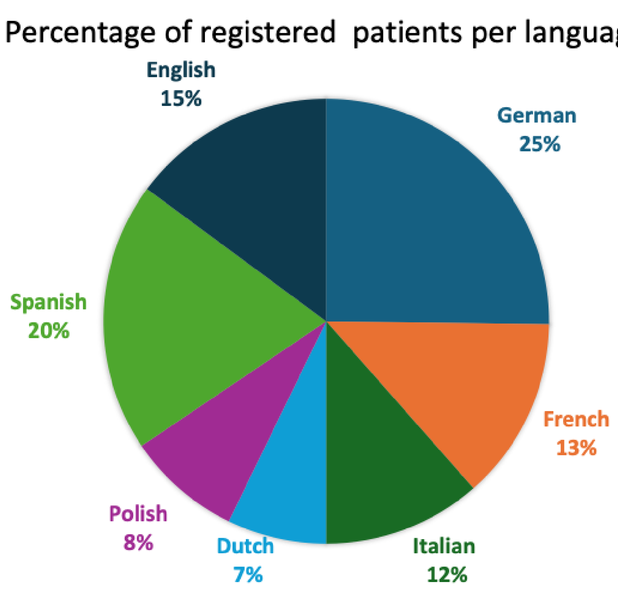
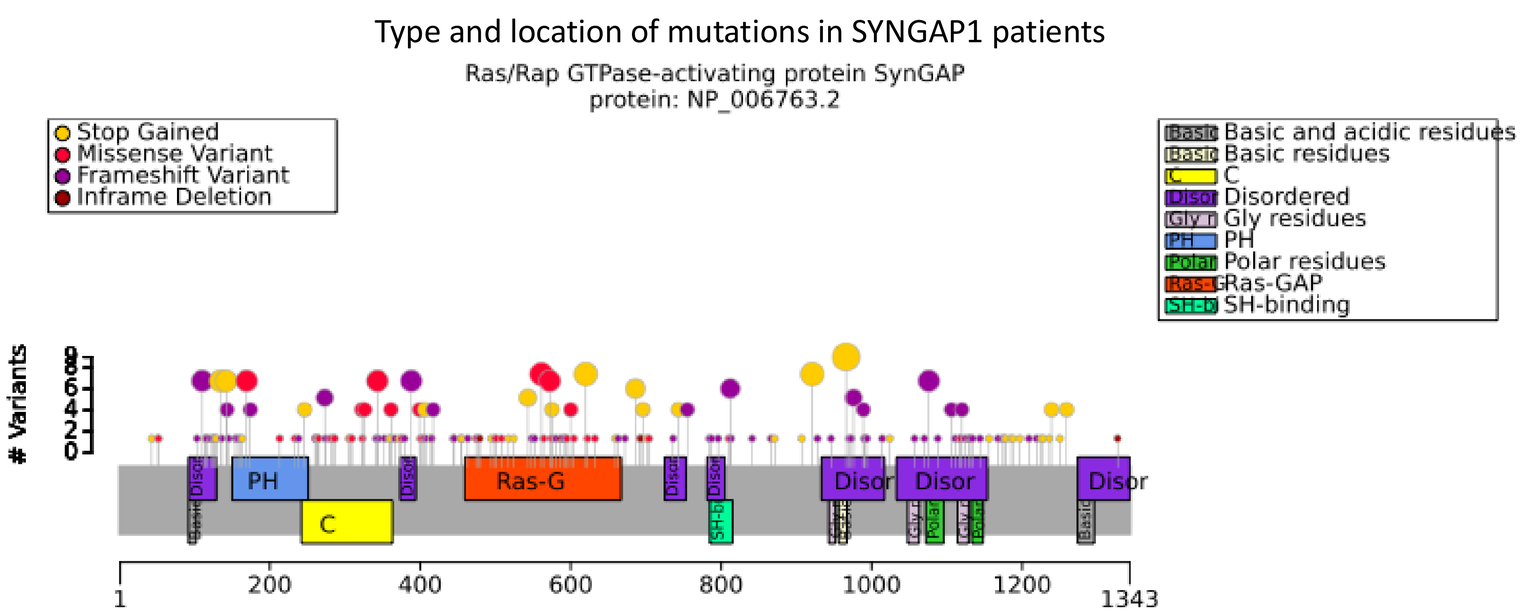
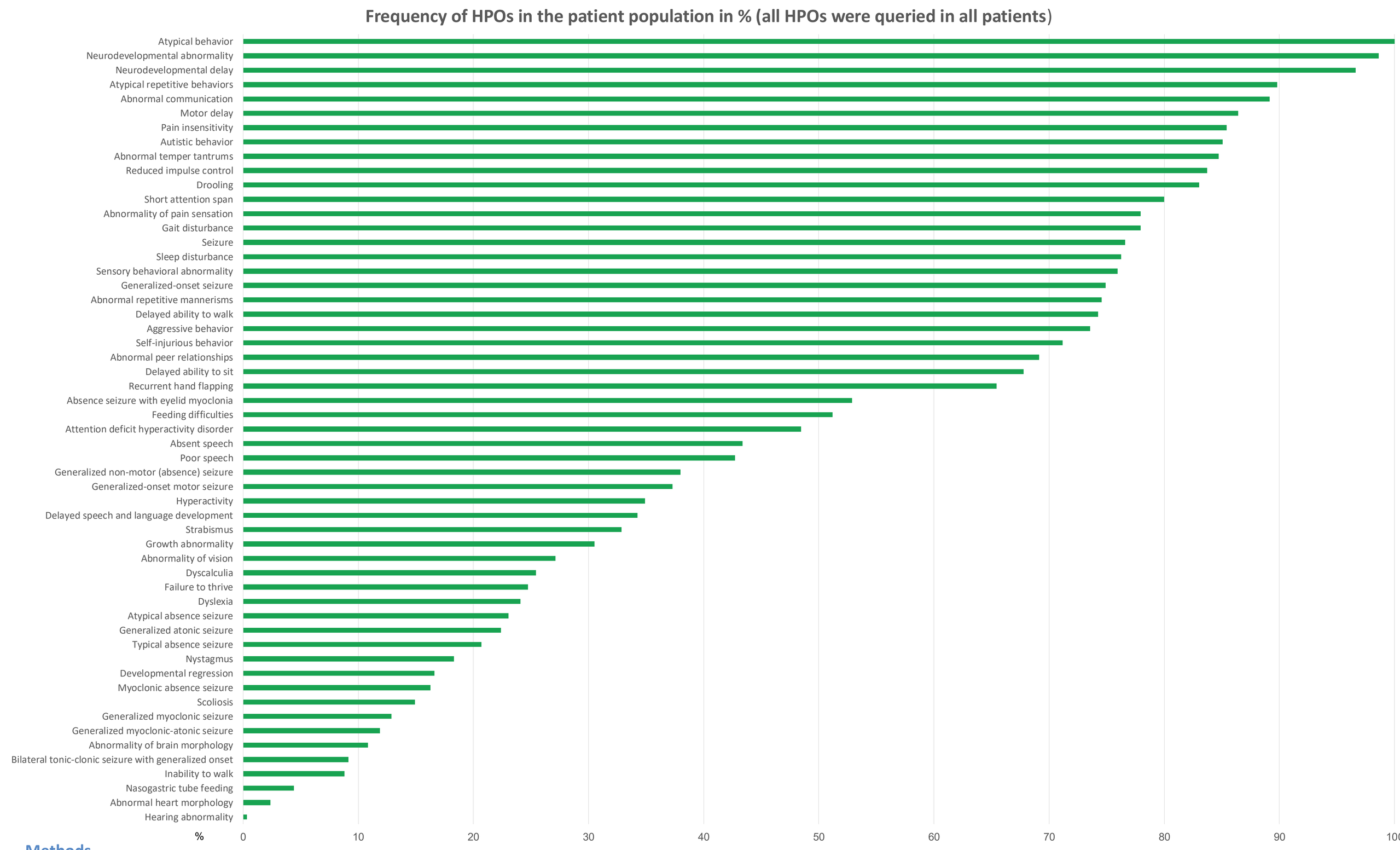
Background

EURAS, The EUropean network for neurodevelopmental RASopathies, aims to accelerate the development of treatments for Cardio-facio-cutaneous syndrome, Costello syndrome, Noonan syndrome and SYNGAP1-related encephalopathy. The Horizon Europe project was initiated by the German SYNGAP1 patient organisation and its medical advisory board.

Through the dedicated PATRE (PATient-Based phenotyping and evaluation of therapy for Rare Epilepsies, www.patre.info) SYNGAP1 registry, the project collects comprehensive health data as a basis for preclinical models. This registry is accessible through a multilingual mobile app to maximize global participation.







Methods

The content of the registry is developed together with patient representatives and is complemented by standardized scores. A domain-specific set of common data elements (DCDEs) based on HPO terms was defined (Kiwull et al., Epilepsia 2024; 65:310–311): A multidisciplinary team of clinicians and patient representatives first defined key clinical topics and selected 40 HPO terms, then expanded this list using bioinformatics tools, and finally reached a consensus on 65 terms through a three-step Delphi process.

Results

A first analysis of HPO freqency is shown above. An exemplary analysis of the development items showed that children with SYNGAP1 experience delayed milestones: **sitting unsupported is typically achieved at 12 months, walking independently at 28 months, and forming two-word sentences at 47 months**.

An exemplary analysis of the **PEDI-CAT score** showed that SYNGAP1 children are on average at the **4th percentile in the "daily activities" domain, at the 12th percentile in the "mobility" domain, at the 4th percentile in the "social/cognitive" domain and at the 9th percentile in the "responsibility" domain** compared to their respective age norm.

Conclusion

The SYNGAP1 registry demonstrates the feasibility and value of a patient-centered, app-based data collection approach to gather insights into rare disorders. The robust, multilingual participation underscores the utility of the registry as a tool for patient engagement and data generation. This platform can serve as a model for other rare diseases, contributing to a broader understanding and support for families worldwide.

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