

ASXL Care Directory: Building the network of healthcare providers for ASXL-related disorders

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Background

The ASXL Rare Research Endowment (ARRE) Foundation is a family-led patient advocacy organization with the mission to improve the quality of life for individuals living with ASXL-related disorders, ultrarare neurodevelopmental disorders caused by pathogenic variants in one of the ASXL genes. Common clinical features include developmental delay, intellectual disability, absent or limited speech, seizures, hypotonia, feeding difficulties, severe constipation, and self-injury. It is thought that approximately 500 individuals are diagnosed and that many more remain undiagnosed.

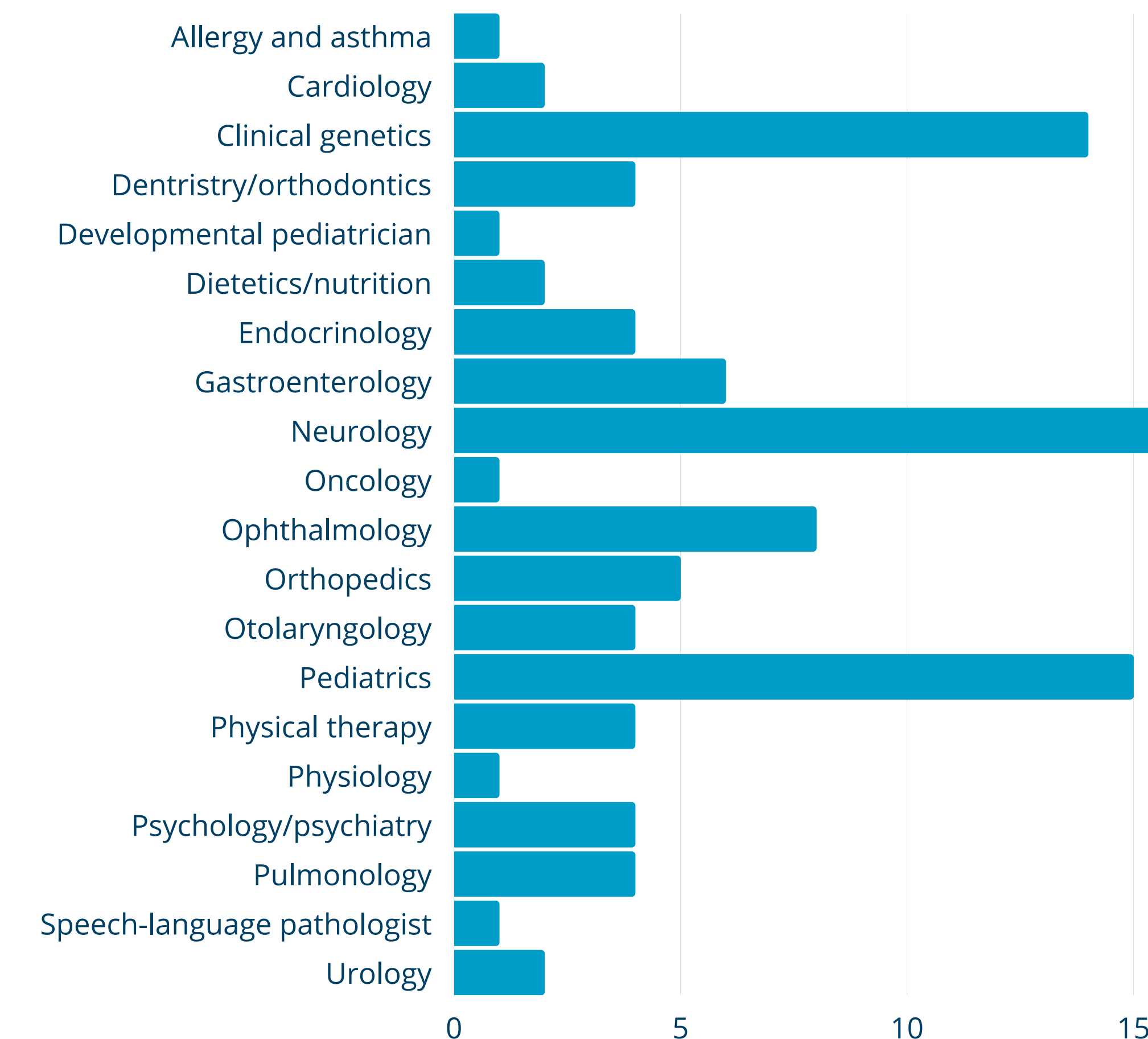
While research interest in ASXL-related disorders is growing rapidly, there remains little understanding of the natural history or mechanisms of disease. Most clinical providers only see one or two patients due to the rarity of these disorders and there are no known treatments or standards of care. The variability and range of the spectrum of symptoms is not well documented and poses additional challenges for research into these conditions.

Purpose and goals

In early 2022, the ARRE Foundation began development of a strategic plan to fill the many research gaps in ASXL-related disorders. One of the priorities identified was to grow the clinical network of specialists who are treating individuals with ASXL-related disorders to provide families with experienced clinicians and build clinical research interest for future studies.

- 1 Short term: Develop list of clinicians to recommend to families**
- 2 Mid term: Build ASXL expertise by referring families to same providers**
- 3 Long term: Develop standards of care using ASXL expert clinicians**

Providers by specialty



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clinicians identified who are seeing multiple ASXL patients



Methodology

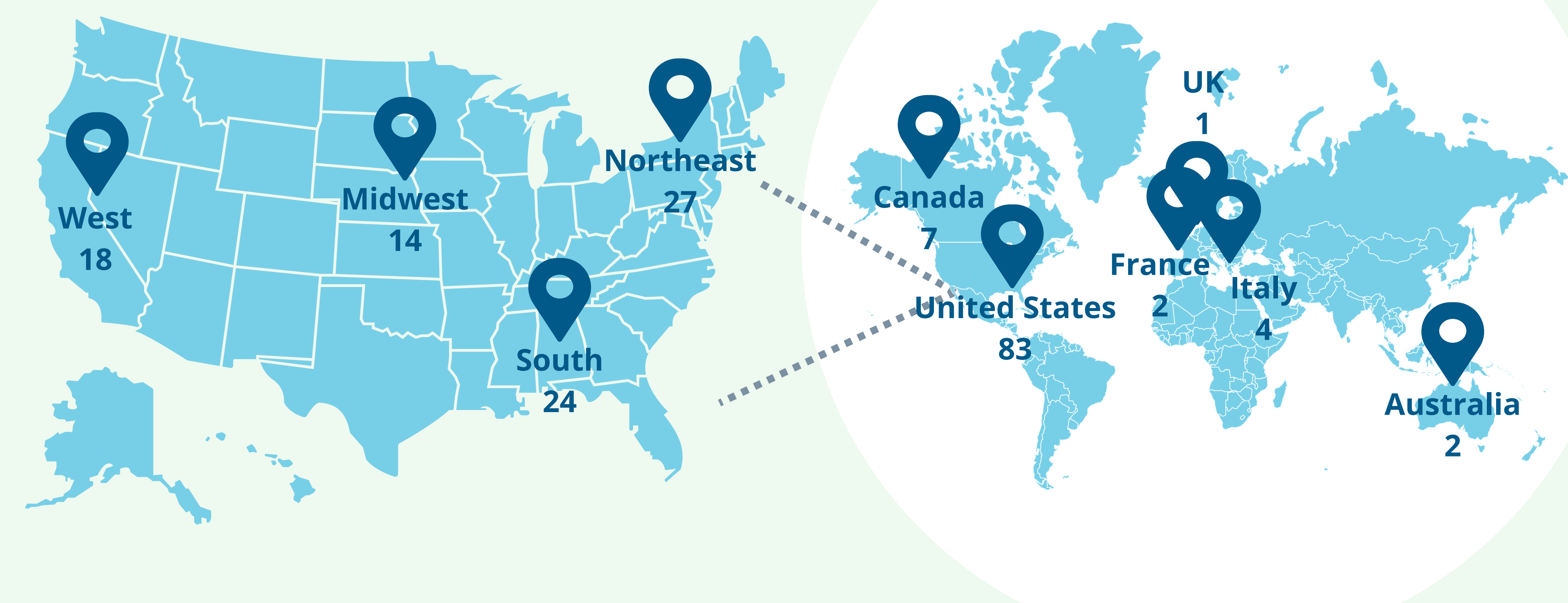
The ARRE Foundation developed the ASXL Care Directory as a first step to identify healthcare providers who are treating individuals with ASXL-related disorders. This is an online directory of doctors, therapists, and other medical professionals. Providers are recommended by families of individuals living with ASXL-related disorders.

To collect family recommendations, we developed an online survey tool (SurveyMonkey) to allow families to submit the name, specialty, hospital/institution, and geographic location of the provider. Given that many individuals with ASXL-related disorders see many specialists, families can submit up to 15 care providers at a time. Guidance provided to families includes suggesting they include healthcare providers who they would recommend to other families, and recognition that providers do not need to have extensive experience treating individuals with ASXL-related disorders but should be a trusted member of their care team.

The ASXL Care Directory is accessible via the ARRE Foundation website. The directory tool is a low-cost application purchased through Elfsight that plugs into the ARRE Foundation website, which is built on the SquareSpace platform. The directory can be filtered on fields we specify, which includes geographic region and medical specialty.

The directory launched to the family community on 2022 Rare Disease Day. Prior to launch, we approached several highly involved families to submit their recommended care providers to pre-populated the directory; when it launched, families could see how it worked. Promotion of the directory, which includes using it and contributing to it, included promotion via social media, mass email, and personal outreach. The ASXL Care Directory is also incorporated into the specific resources we share with newly diagnosed families.

Providers by location



Results

The ASXL Care Directory contains 99 healthcare providers in 21 specialties recommended by ASXL families. The majority of the providers are in clinical genetics (N=14) and neurology (N=16). Providers are included from 6 countries; the majority are from the United States (N=83). 24 families have submitted care providers via the SurveyMonkey form.

Soliciting providers resulted in discovery of three clinicians who were previously unknown to us who are seeing more than one patient with ASXL-related disorders. Those clinicians have joined our ASXL Research Network and have been invited to contribute to a study that includes clinician interviews.

Conclusion

The ASXL Care Directory is a first step in building a larger network of healthcare providers who have experience treating and managing ASXL-related disorders. It is a critical piece of infrastructure for development of future standards of care.

Additional future work includes recruiting additional recommendations for care providers who are located outside the U.S. and who represent additional clinical specialties, and in further engaging identified clinicians in ASXL research activities.