

The rapid deployment of a rare disease community registry

Fillingham, S¹; Henderson, J²; Liu, N², Gwadry-Sridhar, F²

¹PIP-UK, UK; ²Pulse Inframe, USA

INTRODUCTION

Poland Syndrome is a rare birth difference that occurs during foetal development where the baby is born with physical differences. It is estimated to affect 1 in 20,000 newborns and there are currently no effective treatments.

PIP-UK is a registered charity in England and Wales that was established to support and the Poland Syndrome community. For the last 10 years PIP-UK has been conceptualizing and planning a patient registry project to overcome the issues of disparate data, globally dispersed.

OBJECTIVES

To answer this challenge, PIP-UK was focused on ensuring that the data were collected in a standardized way, met regulatory requirements and aligned with their vision to build global collaboration, with the technology as a nucleus for patients to convene and share their experiences.

METHODS

PIP-UK started working with Pulse Inframe in 2021 to launch the Poland Syndrome Community Register. Utilising the Pulse Platform, PIP-UK were able to deploy the registry within 4 months with a recruitment target of 100 participants in the first 3 months.

Common data elements to form the central hub for data to be characterized and curated were created. PIP-UK and Pulse addressed the regulatory requirements that would be needed for drug development by ensuring that the data are collected in a platform that adheres to FDA and EMA data standards and further provides confidence to participants that their data are stored safely due to compliance with HIPAA and GDPR.

RESULTS

The registry currently has 147 active participants enrolled into the platform where 48% of them are under the age of 10. The oldest participants are over the age of 80 years old. Over 40% of participants are received a diagnosis of Poland Syndrome from their surgeon, while only 13% received their diagnosis from a GP.

Figure 1. Age at diagnosis

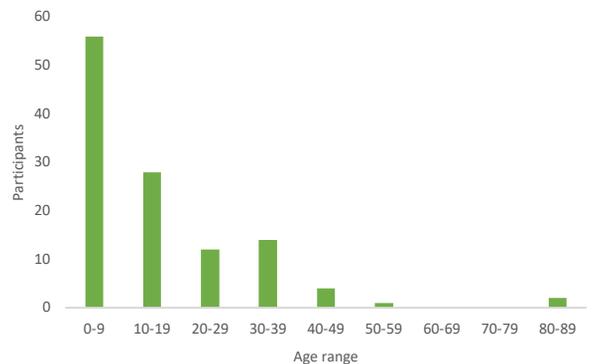
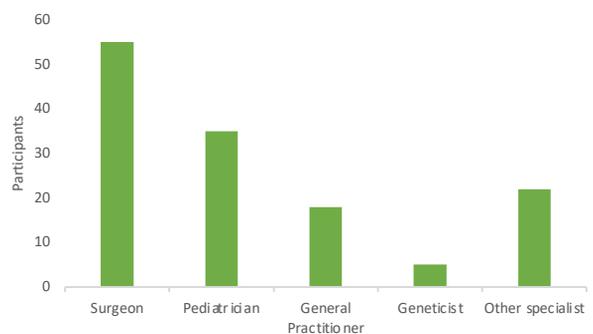


Figure 2. Diagnosis physician



CONCLUSIONS

With advocacy's commitment and leadership PIP-UK has illustrated how a rare disease community can come together from all around the world and collaborate with other stakeholders so that research into their condition can be advanced in using scientific methodology.

