

# The rapid deployment of a rare disease community registry

Fillingham, S<sup>1</sup>; Henderson, J<sup>2</sup>; Liu, N<sup>2</sup>, Gwadry-Sridhar, F<sup>2</sup>

<sup>1</sup>PIP-UK, UK; <sup>2</sup>Pulse 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

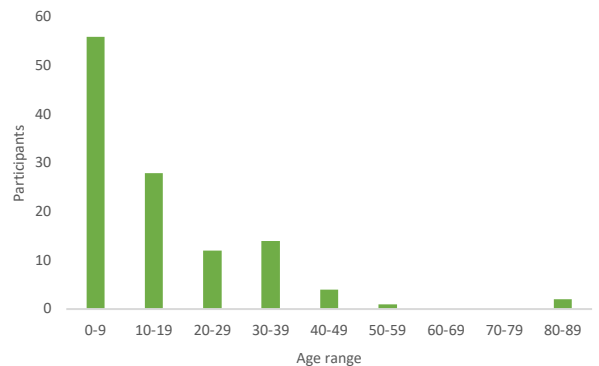
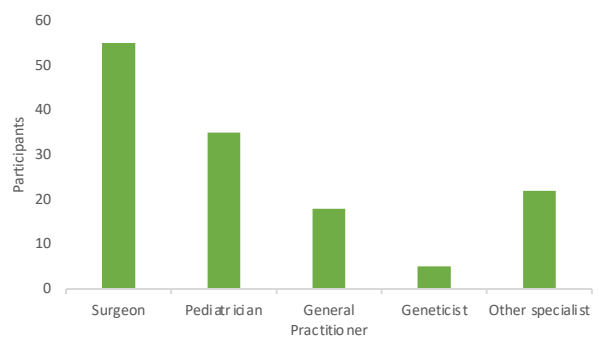


Figure 2. Diagnosis physician



## 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.

