Patient Navigator for The Office for Rare Conditions: A progress update.

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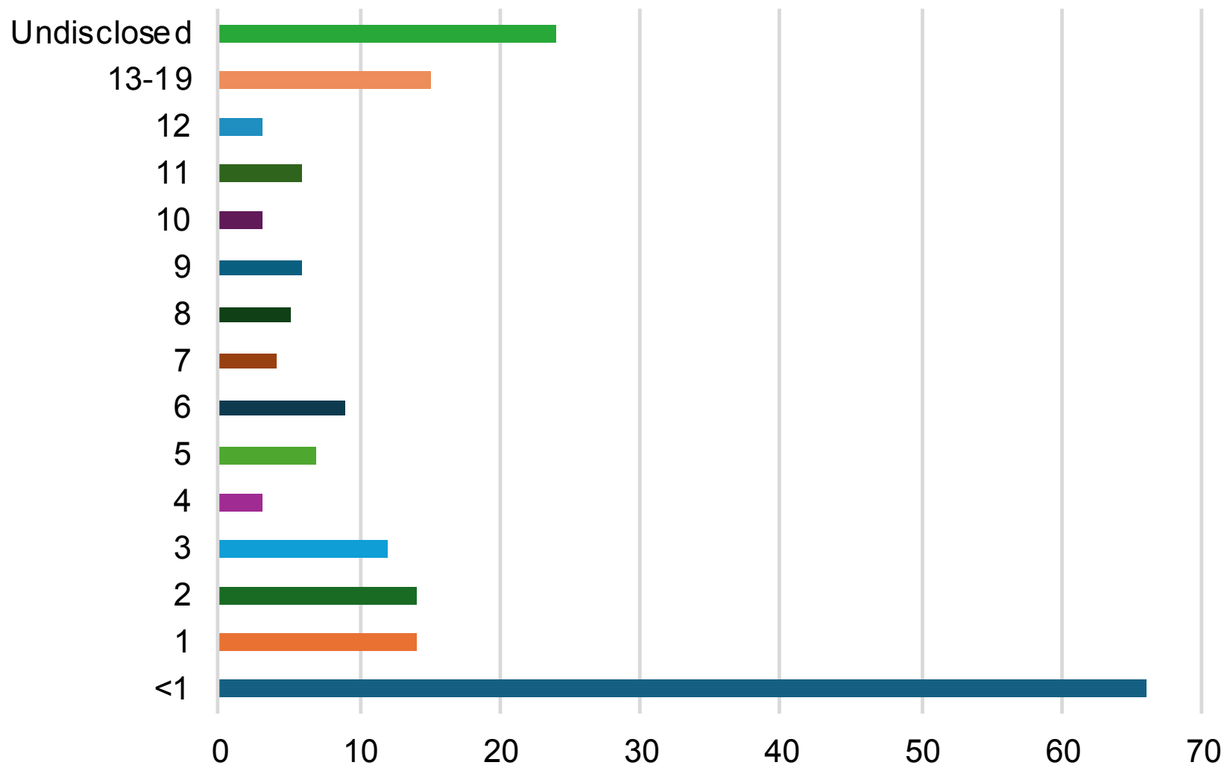
Introduction

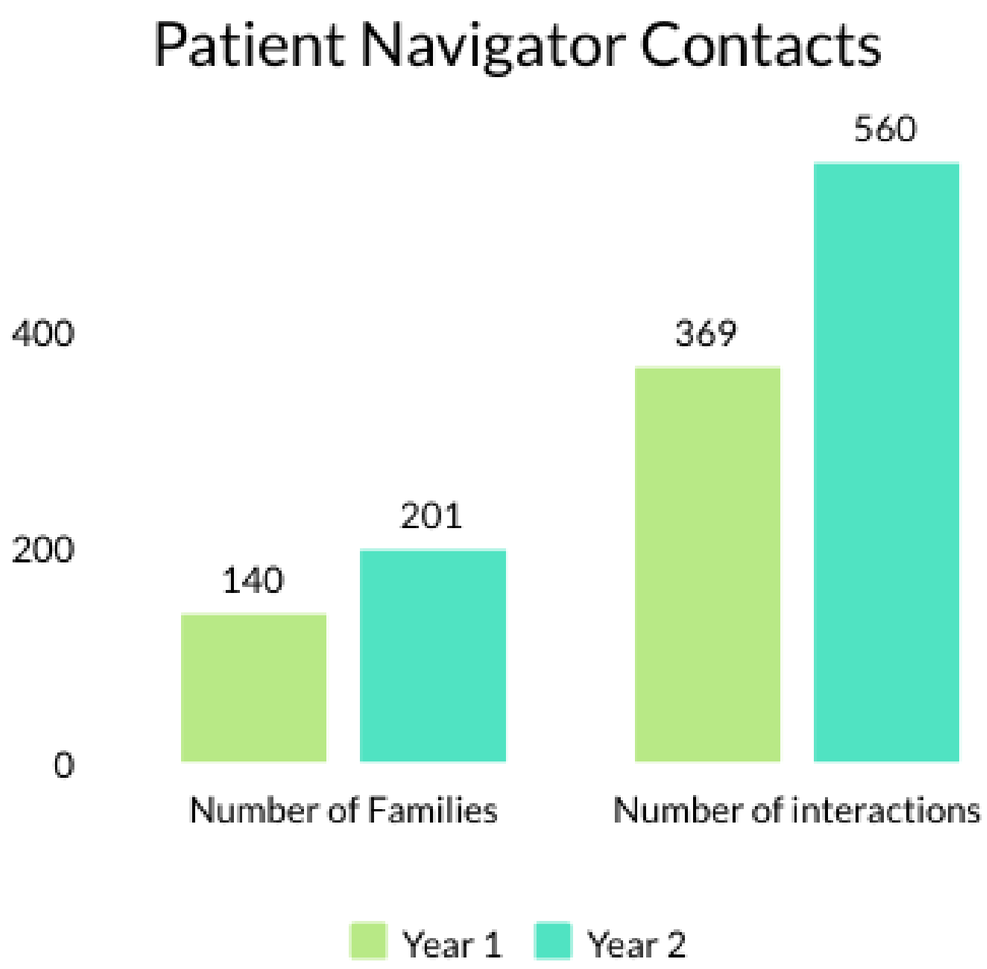
The Patient Navigator role was the first of its kind in Scotland and was established in 2023. The role of the Navigator was to raise awareness, offer support, signposting and education, improve care coordination and provide holistic care to those with rare, low prevalence or undiagnosed conditions. This involved addressing the physical, mental, financial and social needs of families.
We present data from the origin of the role in January 2023 until September 2024.

Age of Patients



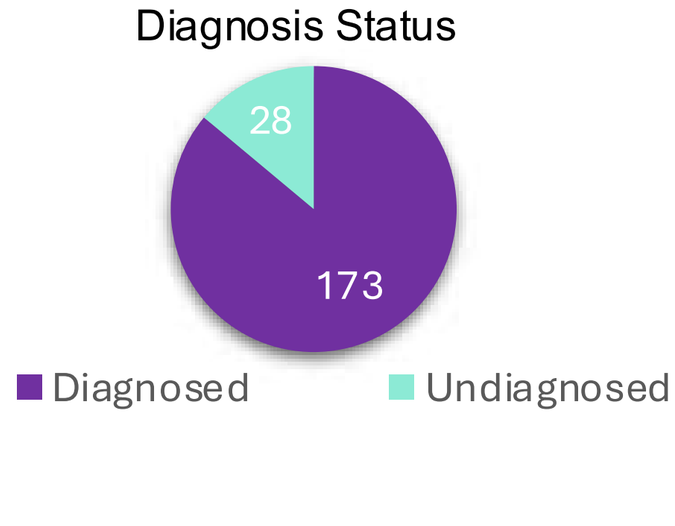
Results

We have shown the number of families the Navigator had contact with, the number of interactions and how this increased from the 1st to the 2nd year of the role. There was a median of 2 interactions with the Navigator.



A range of contact methods were used to suit the needs of the family, including face to face, email and phone. 284 interactions took place via email, making it the most common form of communication.

Proportion of patients with a diagnosis:



Conditions Encountered

134 rare conditions were encountered with prevalence ranging from 1 in 2000 to being only one of a few worldwide. Examples of some of the conditions encountered are shown below.

Conditions Encountered			
Phelan-McDermid Syndrome	Hirschsprung's Disease	Congenital melanocytic naevi	Ehlers Danlos Syndrome
Beckwith Wiedmann Syndrome	Spinal muscular atrophy type 1	Fabry Disease	Ascending hereditary spastic paraplegia
Morgagni-Stewart-Morel Syndrome	Neurofibromatosis type 1	Kabuki Syndrome	William's Syndrome
Duchenne muscular dystrophy	ARPKD	Klinefelter Syndrome	Prader-Willi Syndrome
Pierre Robin sequence	Aicardi-Goutières Syndrome	Myotonic dystrophy type 1	Mowat-Wilson Syndrome
White-Sutton Syndrome	Dysfibrinogenemia	Antiphospholipid Syndrome	Joubert's Syndrome
Simpson-Golabi-Behmel Syndrome	Moyamoya	Aplastic anaemia	Noonan's Syndrome
Laryngo-onycho-cutaneous Syndrome	Angelman Syndrome	Sotos Syndrome	Coffin Siris Syndrome
Patau Syndrome	Klippel-Feil Syndrome	Retinitis Pigmentosa	Poland Syndrome
Goltz Syndrome	Di George Syndrome	Pura Syndrome	Oesophageal atresia

Resources Developed

Resources included a holistic needs assessment form, a healthcare professional contact log and a database detailing resources for support in finance, housing, mental health and wellbeing, activities, charity contacts, refugees and sibling support.

Areas of Support

Families were offered general support, support with funding and education, signposted to support groups and psychological support, and aided in linking with healthcare staff and end of life care.

Feedback

Questionnaires were given out to families, with 96% reporting the information they received was helpful, and 100% stating they knew who to contact if they needed further support.

Conclusion

The Navigator has proved a busy and effective role that has continued to grow and receive excellent feedback. Families often contacted the Navigator feeling lost and helpless and were offered diverse support. This role should be considered by other centres caring for those affected by rare conditions.