

Rare diseases are a significant health problem, often associated with poor outcomes. A rare disease is one that affects 1 in 2000 or fewer patients, with an estimated 170,000 people in Wales living with a rare disease. The Syndrome Without A Name (SWAN) Clinic was implemented to improve the experiences and outcomes of patients with undiagnosed rare diseases in Wales. This is the first known service of its kind in the United Kingdom, and aims to reduce the diagnostic odyssey, increase the diagnostic rate, and improve care coordination for patients. An evaluation of the SWAN Clinic pilot was carried out to assess and measure the impact of the service on service users' experience and outcomes, and to identify the factors affecting implementation success. Evaluation methods included surveys of service users and their families, interviews with service users and staff, and routinely collected clinical data.

Service users' experience of the SWAN Clinic was overwhelmingly positive. They appreciated the multidisciplinary and holistic nature of the clinic, and commented on how this was a stark contrast to their previous experiences of care.

Improvements in service users' self-management capabilities were observed between the point of referral to the SWAN Clinic and 6 months after the initial clinic visit. Data on patients' perceived control of their condition, and patient enablement as a result of the SWAN Clinic, were collected. However, these were not sufficient to draw robust conclusions.

A number of unanticipated barriers to the implementation and delivery of the clinic were identified via the evaluation. These were largely related to delays with implementing genetic investigations in the clinic, which, to date, have prevented necessary investigations from taking place for many patients. A number of additional barriers and facilitators to the implementation of the SWAN Clinic were also identified in the evaluation related to accessibility, resource availability, infrastructure and governance.

To date, of those seen in the SWAN clinic, a diagnosis has been provided to 12% of all adult patients, and 7% of paediatric patients. However, for those patients who have undergone the appropriate investigations and been discharged from the service, the diagnostic rate increases substantially to over 61%. This suggests that while the aim to increase the diagnostic rate to 40% of referrals is not yet fully realised, once the barriers to implementation have been overcome, the SWAN Clinic has the potential to meet this target. This would significantly improve care for patients with undiagnosed rare diseases in Wales and realise its aim of reducing the diagnostic odyssey by providing patients with a definitive and timely diagnosis.

Furthermore, the SWAN Clinic provided a number of key benefits for service users and their families. In addition to those described above, the improved care

coordination provided by the clinic was a major benefit. A number of patients have also had substantial changes made to their treatment or management as a result of their referral to the clinic, despite not yet having a diagnosis. Moreover, in some cases, even when a definitive diagnosis was not available, some potential diagnoses could be ruled out, which patients also found reassuring, indicating a clear benefit to patients.

This pilot study demonstrates that patients are benefiting from a more holistic and coordinated approach to their care, compared to that provided prior to their referral into the SWAN Clinic. This evaluation also identified some strengths and weaknesses in its infrastructure and service provision, which the clinic can target for further improvement.