Tales from the SWAN Clinic

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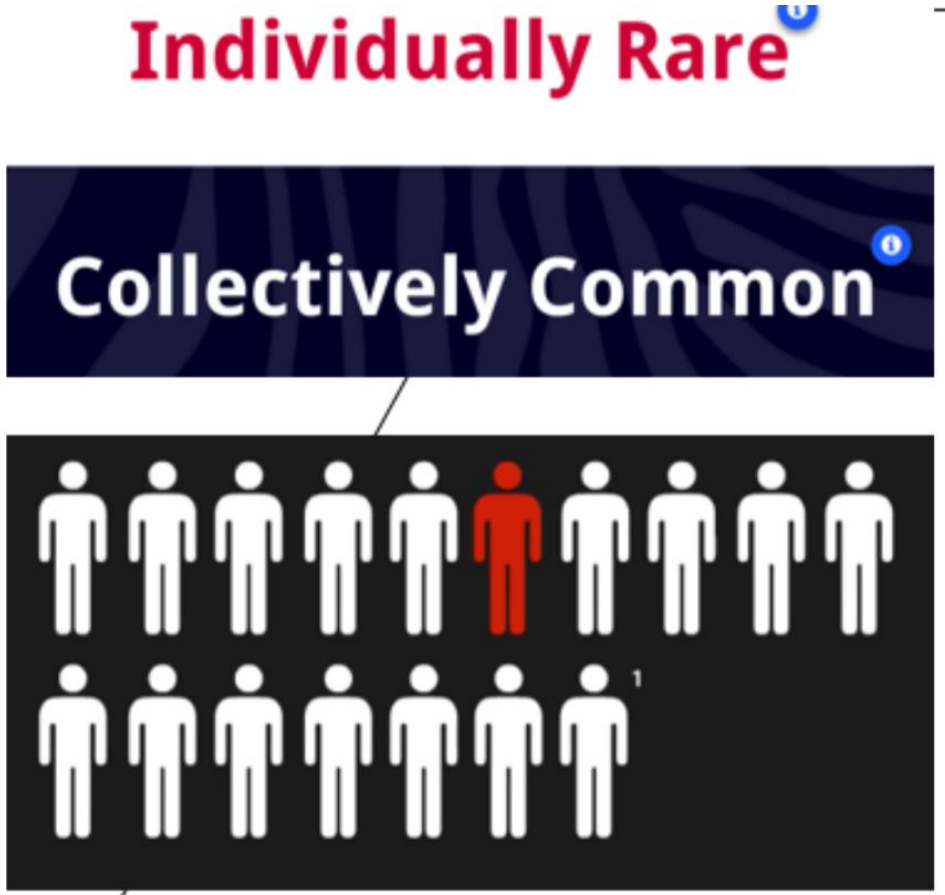
Content

- SWAN Clinic – what is it?
- Rare Disease Strategy and purpose
- Outcomes
- Some SWAN Tales
- Conclusions

RARE DISEASES

- A disease affecting < 1 in 2000 people = RARE DISEASE
- Over 7000 RARE DISEASES described
- Estimation of 3.5 million people in UK have rare disease (170,000 people in Wales)
- **1 in 17 people have a lifetime risk of being diagnosed with a rare disease**

- 80 % present in childhood
- 30 % of those die before age 5
- 70-80% suspected genetic origin



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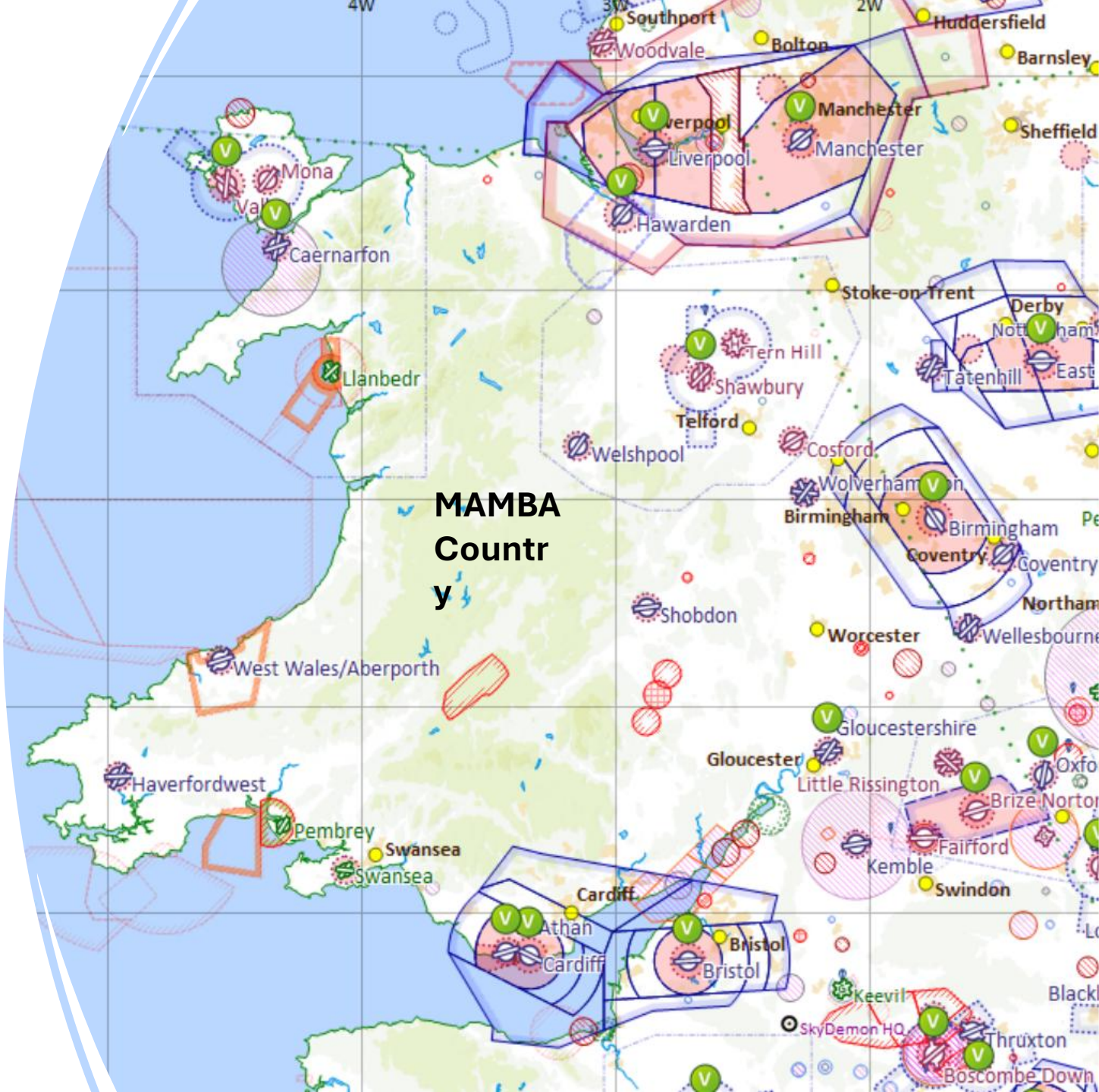
Matt Spencer

SWAN a National Service for Wales



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SWAN MDT

SWAN Lead

Paediatric Team

- Paediatric clinical lead
- Paediatric Genetic lead
- Rare Disease Paediatric Clinical Fellow
- Paediatric SWAN Clinical Nurse Specialist

Adult Team

- Adult clinical lead
- Adult Genetic Lead
- Rare Disease Adult Clinical Fellow
- Adult SWAN Clinical Nurse Specialist

Additional Team Members

- Genetic Counsellor
- Bioinformatician

SWAN Clinic Referral Criteria

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- Patients who are referred to this clinic should usually have the involvement of two or more systems
- Consultant initiating the referral highly suspects a unifying underlying diagnosis.
- They may have congenital malformations or dysmorphism suggestive of an underlying monogenic disorder
- Supportive family history
- It is expected that reasonable investigations, including genetic testing where indicated, without identifying a diagnosis have been completed (and are not pending).
- Referrals from secondary care (though Primary Care referrals considered on a case by case basis)
- SWAN clinic does not take over the care of the patient from the referring team

SWAN MDT and Clinics

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- Weekly Joint SWAN MDT (Adult and Paed)
- If Adult or Paed workload requires it – MDTs can be run separately
- Business and Clinical components
- Re-analysis MDT – Bioinformatician led
- Metabolomics MDT
- Adhoc MDTs with specific specialties
- Fortnightly clinics – Adult and Paed (2-3 patients per clinic)
- 3 monthly link in meetings with UDNI and Perth Rare Disease Centre (and others eg Helsinki)

Clinical Fellow Role

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- 50% (5 sessions a week) – Monday, Tuesday, Alternate Wednesdays
- Present referrals with brief history
- Accepted patients worked up and “Patient Report” produced
  - All appointments and letters
  - Investigations list
  - Contact specialties
  - Link between genetics and other medical specialties
- Chair weekly MDT
- Arrange on going investigations
- Work closely aside Clinical Nurse Specialists

Dr Matthew Spencer

Rare Disease Paediatric Clinical Fellow

Dr Aung Saw

Rare Disease Adult Clinical Fellow

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Siân Williams

Adult SWAN Clinical Nurse Specialist

Zoe Morrison

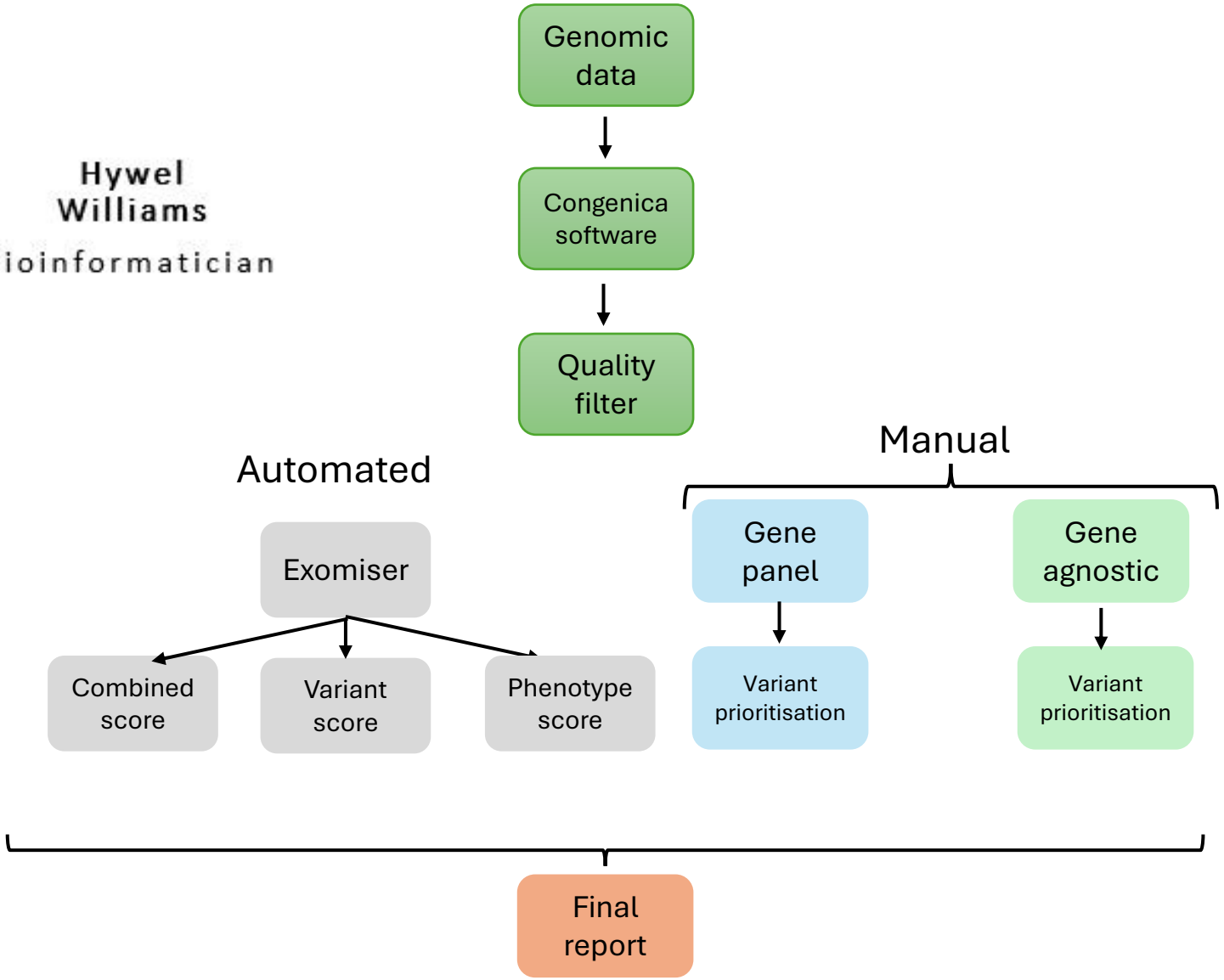
Paediatric SWAN Clinical Nurse Specialist

Role of the Clinical Nurse Specialist

Re-analysis Pipeline

Hywel Williams

Bioinformatician



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SWAN Overview

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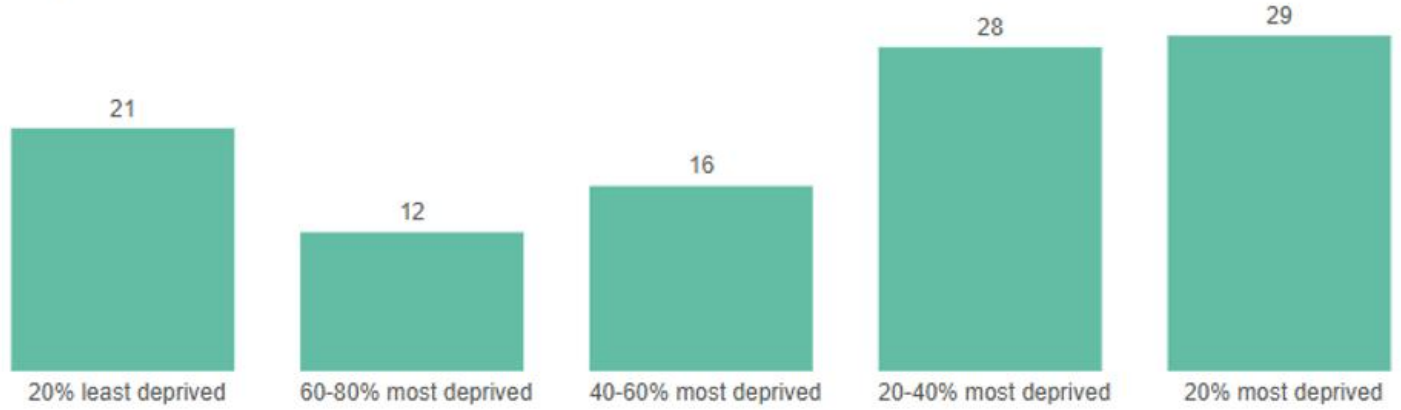
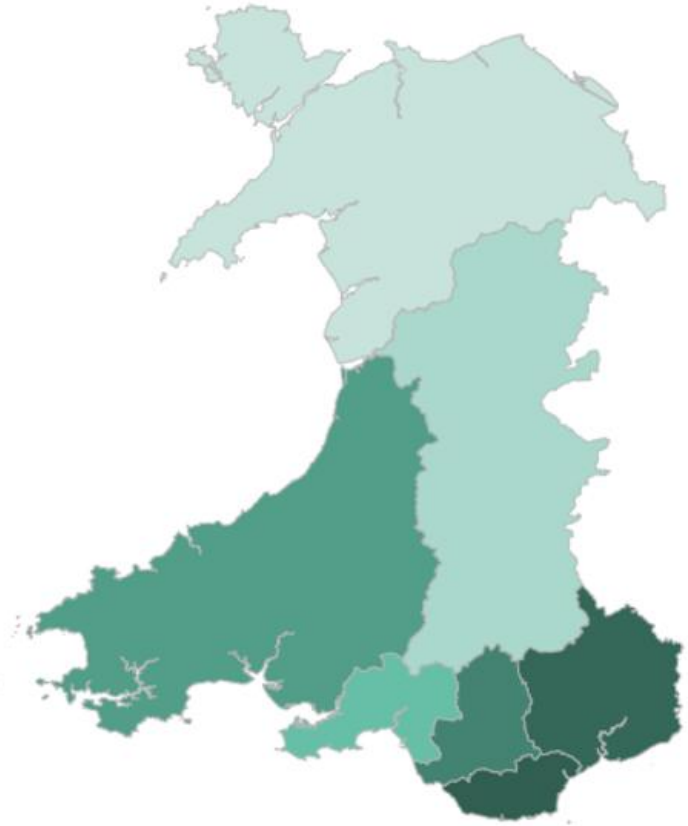
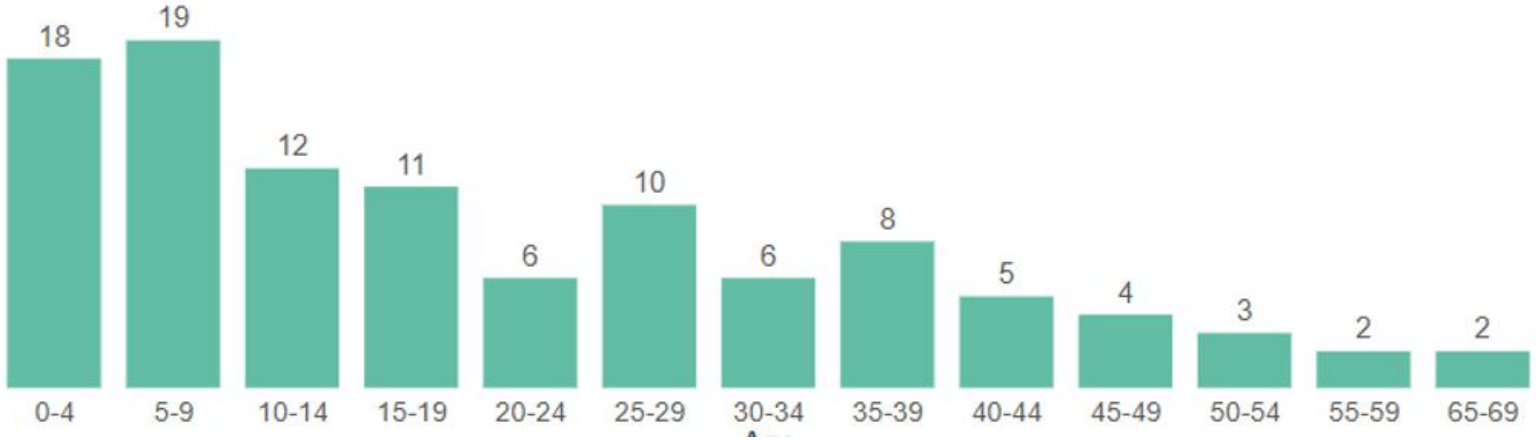
Patients by Deprivation Quintile



Residence Health Board



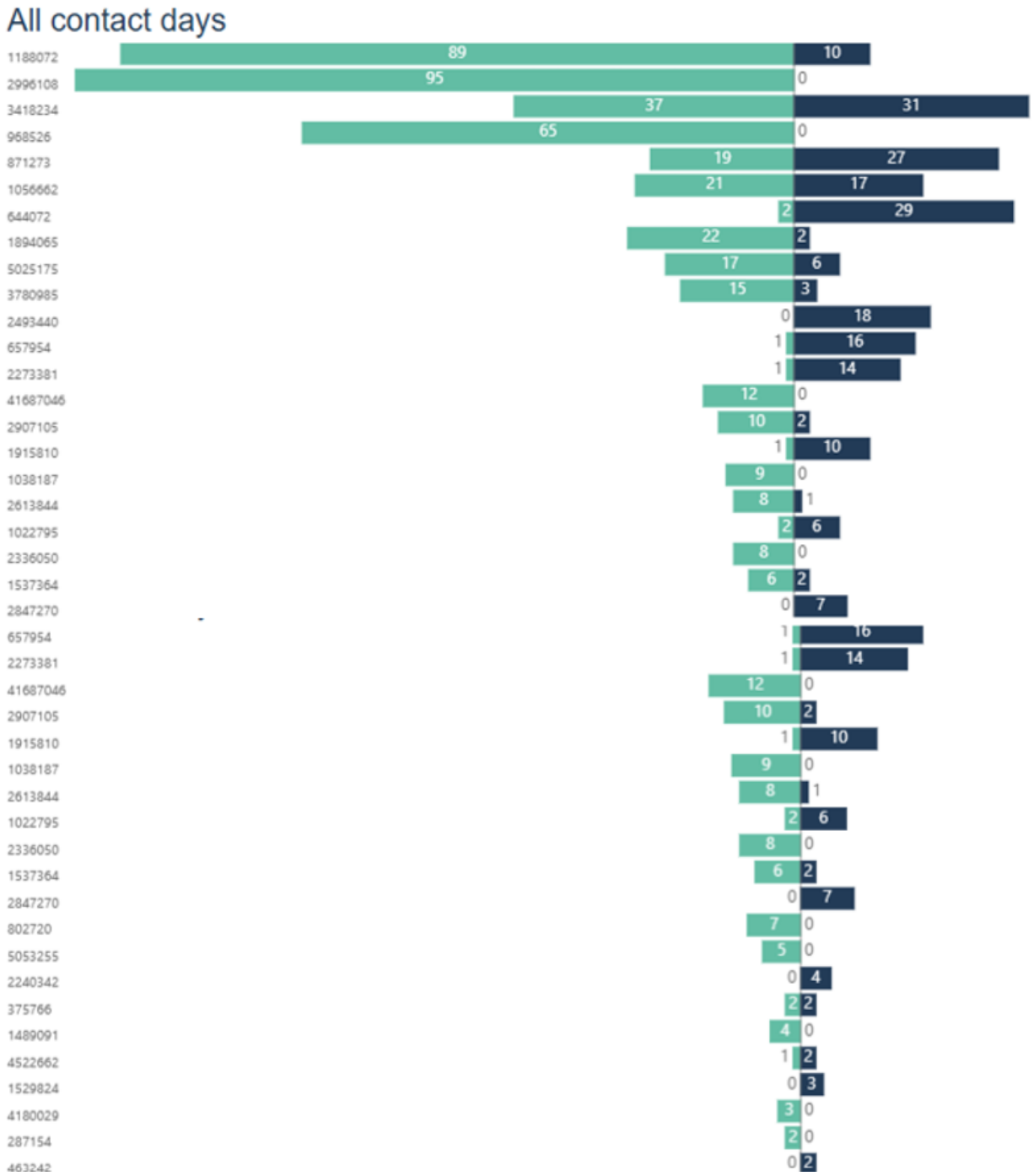
Patients by Age



SWAN Outcomes

Contact Days – OPD, Inpatient, Emergency Department 6 months before and 6 months after

Contact days for SWAN patients was 23x the expected mean compared with the standardised national rates before and 10x after



SWAN Diagnostic Rate

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- Paediatric rate – confirmed, partial or likely diagnosis (minus withdrew consent) – 19%
- Paediatric confirmed only – 12.7%
- Adult rate – confirmed, partial or likely diagnosis (minus withdrew consent) – 31%
- Adult confirmed only 12%

Challenges for SWAN

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- Secure ongoing funding
- An important but delicate balance with Genetics
- How to reach out across geography (MAMBA country)
- Clinical support but vitally also laboratory support and send support
- Paediatric vs Adult cases (need to have equal access to diagnostics for adults)
- Referral sources and selection of cases
- Specialist advice – individualised MDTs
- Staff churn as expertise needs to be gained
- Biobank and consent issues for deceased
- Training – due to the novelty this doesn't easily sit within current training programs

Who needs to be in the room

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- Clinicians
- Treaters prepared to reach out for compassionate use or ipfr
- Genetics
- Bioinformaticians
- Nursing
- Other specialists as required
- AI

Pushing the boundaries of rare disease diagnostics with the help of the first Undiagnosed Hackathon

Angelica Maria Delgado-Vega, Helene Cederroth, Fulya Taylan, Katja Ekholm, Marlene Ek, Håkan Thonberg, Anders Jemt, Daniel Nilsson, Jesper Eisfeldt, Kristine Bilgrav Saether, Ida Höijer, Ozlem Akgun-Dogan, Yui Asano, Tahsin Stefan Barakat, Dominyka Batkovskyte, Gareth Baynam, Olaf Bodamer, Wanna Chetruengchai, Pádraic Corcoran, Madeline Couse, Daniel Danis, German Demidov, Eisuke Dohi, Mattias Erhardsson, ... Ann Nordgren + Show authors

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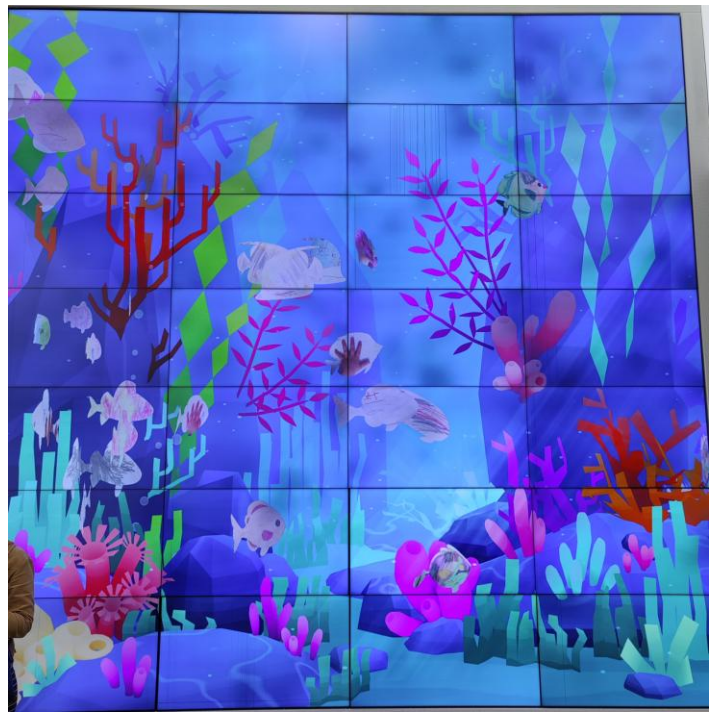
Importance of being connected to other specialties, labs, research, pharma, initiatives and friends

Patients with rare diseases diagnosed after international hackathon

Researchers, doctors, molecular biologists and data experts gathered on June 17–18 to crack unsolved disease diagnoses. Photo: Fredrik Persson

Collaborations

- Regular **meetings within** Undiagnosed Diseases Network
  - Perth, Australia
  - NIH, Harvard, USA
- Sharing experience for those looking to offer SWAN services
  - London, Birmingham, Ireland
  - Sydney, Australia
- International Conferences – Nursing, Genetic, Specialist
- Linking with Helsinki Rare Disease Centre
- Baylor, CHOP, Toronto Childrens, Kings, Leeds, GOSH
- Undiagnosed Hackathon 2023/24



Conclusions

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- It has been a privilege to be involved in the SWAN clinic
- The clinic offers the precious gift of time
- It has been possible to extend diagnostic testing and its yield
- Not everything is genetics
- Holistic coordination of care role is of critical importance to families
- Targeted treatment is enabled with the MDT supporting IPFR or compassionate use
- Colleagues around the world and locally have given of their time, advice and testing with extraordinary generosity in the interest of patient care – thank you!


Thank you