Why weren’t we told?
= Implementation of a Pre-conception Genetic carrier screening program

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- We are all carriers of at least one lethal genetic condition
- 1 in 20 people in Australia are carriers for either cystic fibrosis, spinal muscular atrophy or Fragile X syndrome¹
- In July 2018 the Royal Australian College of Obstetricians and Gynaecologists changed its position statement on genetic carrier screening to advise that “Information on carrier screening for the more common genetic conditions that affect children (e.g. cystic fibrosis, spinal muscular atrophy, fragile X syndrome) should be offered to all women planning a pregnancy or in the first trimester of pregnancy”.²

Problem: Genetic Carrier screening is not routinely offered to patients of reproductive age attending the genetic clinic.

SMART Aim: Patients attending the genetic clinic who are of reproductive age and planning a family, or in early pregnancy (<12 weeks) will be offered genetic carrier screening. How much: 100% When: by December 2018

Priority change ideas:
- Develop a consensus screening protocol
  Who: all staff met to brainstorm ideas
- Send patients information on carrier screening
  Who: admin staff
  When: at time of sending clinic appointment details
- Commence discussions on carrier screening
  Who: clinical staff
  When: in clinic appointment
  Whom: All patients of reproductive age planning a pregnancy or in first trimester of pregnancy.

Results

Patient response to the project:
- Were you aware of carrier testing before coming to our service? No (7/9) Strongly agree/agree (7/9)
- Did you read the information brochure? Yes (8/9) Strongly agree/agree (7/9)
- It was appropriate that carrier screening was discussed in the appointment. Strongly agree/agree (7/9) Disagree (4/7)

Project Outcome:
- Overall, 40% of eligible patients were offered carrier screening.
- Response rate for staff surveys was only 40%.
- Patient feedback was generally positive but again, response rate from the patient survey was low.

Lessons learned:
- We succeeded in raising awareness of carrier screening amongst our patients. Many patients indicated that they planned to discuss carrier screening with their friends/family.
- We succeeded in developing a screening protocol and process that was user friendly and that could be used in a busy genetic clinic.
- The project leader is integral in engaging staff to maintain change.

Acknowledgements:
- Dept of Clinical Genetics RNSH
- Patients of the Genetic Clinic
- CEC

References:
1. Archibald et al Genetics in Medicine 2018;20:513-523
2. RANZCOG Statement on “Prenatal screening and diagnosis of chromosomal and genetic abnormalities in the fetus in pregnancy.” C-Obst 59 August 2018.

Moving forward: Ultimately, carrier screening will be more effective if implemented by primary care physicians. Clinical genetic professionals can assist in providing expert advice and helping to develop processes and resources for GP’s etc.