Genetic Counselling framework

Section A – exploring with individual/couple their opinions and thoughts of having a family knowing about their gene variant. Points to consider include:

- Explore what has brought the couple to seek information.
- What is their starting point/ understanding/ feelings about the situation?
- Unpack their understanding of their family history and their perception of risk of
  a) passing on a gene variant and
  b) implications to future children of inheriting the gene variant.
- Balance couple’s understanding with current knowledge about gene penetrance, age of onset, range of cancers etc.
- Unpack the impact on the person who has the gene variant- is their experience shaping their desire not to have a child with the same variant?
- What is it like for them?
- Is their experience unusual? Can GC give perspective to their experience? E.g site within usual observed range of experience? What makes their experience unusual? Is their experience likely to be repeated?
- Is the motivation to explore reproductive options shared equally by the couple? Is it one-sided? Are there other family members, or significant others who have influence over their decisions? Explore where the motivation is coming from and unpack.
- Explore the potential impact on them as parents if they had/didn’t have a child with the gene variant.
- Thought experiment re. future treatments, improvement in care, how would that impact on their views now?
- Seek to arrive at a shared honest understanding between and with the couple about what is known and what is not known about their familial gene variant’s penetrance contextualised in their family history and what that means to them.

Section B – information giving in relation to all available options:

Information giving about all possible options should be part of the discussion with all couples/individuals, options to discuss:

1. Pre-natal diagnosis (PND),
2. Pre-implantation genetic testing for monogenic conditions (PGT-M),
3. Natural conception without intervention, and
4. Bespoke non-invasive prenatal diagnosis (NIPD) if pathogenic gene variant would be paternally inherited.
5. Adoption
6. Gamete donation (egg and/or sperm donation with assisted conception)

- Remember to mention to individuals/couples during conversation that options are available and that it is not a recommendation. Important to acknowledge patients may feel obligated to choose some form of intervention because the option is being given by a health professional. Explain that this is not the case and that we will support the individual/couples choice.
Likely individual or couple will fit into one of the below categories:

➢ Wish to have a child without any intervention
➢ Would like to avoid passing on the pathogenic gene variant (given understanding of condition and residual risk) and would not want to intervene in a pregnancy.
➢ Would like to avoid passing on the pathogenic gene variant (given understanding of condition and residual risk) and would consider intervening/ ending an affected pregnancy.
➢ Would like to avoid passing on the pathogenic gene variant and would consider options for having children who are not biologically related.

If individual/couple wish to have a child without any intervention - structure of conversation could follow:

• Support their decision, ensure they are aware of the inheritance pattern and timing of appropriate genetic testing /screening for their future children.

If individual/couple wish to avoid passing on gene variant - structure of conversation could follow:

• Would they be comfortable with ending an affected pregnancy?
  a. If no – only option for biological child is PGT-M. If considering a non-biological child, options include adoption and gamete donation (sperm or egg donation and assisted conception)
  b. If yes – all above options available

• PGT-M discussion to include:
  a. Timing of PGT-M – a long process from referral to embryo transfer. Around 12 months but could be longer or shorter depending on factors like specific condition and family structure.
  b. DNA samples and consent for use of samples for PGT-M very likely to be needed from individual/couple and affected partner’s affected parent.
  c. Process includes IVF which can be emotionally and physically difficult for both the individual/couple.
  d. Elements of PGT-M can be described as invasive for partner undergoing ovarian stimulation.
  e. HFEA licence: If no HFEA licence granted, one can be applied for and referral to PGT-M service should still be made if PGT-M is the individual’s/couple’s preferred route.
     i. Please note there is a chance that PGT-M might not be available if HFEA licence is rejected, important to manage patient expectations around this.
     ii. Likely to be a delay of a number of months to PGT-M process whilst licence is being applied for.
  f. For NHS funded PGT-M, couple need to fit specific criteria. Self-funding and private options may be available if criteria not met.
  g. PGT-M using a sperm donor, egg donor or surrogate is more complicated and only partially funded if eligible for NHS funded PGT-M.

• PND discussion to include:
**PND is only an option if couple would end an affected pregnancy and should not be used as a predictive genetic test for a child**

a. Invasive testing – CVS/Amnio  
b. Non-invasive prenatal testing - paternal allele exclusion (not an option if condition would be maternally inherited)  
c. Fetal sexing (non-invasive) and invasive testing (CVS/Amino) if gender specific variable penetrance cancer susceptibility condition.  
   o Important to ask whether sex of pregnancy would impact on decision-making – if couple would want to end an affected pregnancy regardless of sex – fetal sexing not indicated.

- Gamete donation discussion to include:  
  a. Egg donation - unlikely to be able to access funding for this. If a same sex male couple – would also need a surrogate  
  b. Sperm donation – unlikely to be able to access funding for this.

- Adoption  
  a. Different adoption rules and costs by local region

- Ensure individual/couple are aware they can change their minds or revisit discussion in the future.

**Section C: Discussion – explore the following points as part of a discussion of the above:**

**Congruence**  
- Explore each partner’s individual thoughts and feelings about the options available to them  
- Do they agree? If not, facilitate discussion of the areas they view differently  
- Allow time, encourage further discussion between them outside of the counselling setting, offer follow-up if appropriate

**Decision regret**  
- Support the couple to consider possible future challenges and how they might feel/cope with these, to include:  
  o Having a series of affected pregnancies and terminations  
  o Unsuccessful PGT-M  
- Acknowledge that decisions may change with future experiences and circumstances, reassure that this would be supported

**Support network**  
- Who are the key people in their lives who they will seek support from?  
- Do they think their friends/family will agree with their decision, or might this cause conflict?  
- Are there ethical/religious/cultural factors to consider?

**Timing**  
- Are they ready to start a family now, or making plans for the future?  
- Ensure they understand the pathways and timescales for the options they are considering