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Cross specialty implications of Human Genetics Society of Australasia's EVOLVE list

-- A/Professor Julie McGaughan

evolve
evaluating evidence. enhancing efficiencies.

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Full HGSA EVOLVE list

1. Do not use brain magnetic resonance imagery (MRI) for routine surveillance of asymptomatic neurofibromatosis type 1
2. Do not undertake sequential testing for heterogeneous genetic disorders when targeted next generation sequencing (NGS) is available
- 3. Do not undertake genetic testing for methylenetetrahydrofolate (MTHFR), apolipoprotein E (APOE) and other such tests where the clinical utility for diagnostic purposes is extremely low**
4. Do not undertake carrier state testing for rare recessive disorders where partner has a family history, the couple is non-consanguineous and there are no common causative mutations
5. Do not undertake genetic testing when clinical diagnostic criteria exist and there are no reproductive or predictive testing implications

- There are some conditions where the presence of particular allele(s) is neither necessary nor sufficient to cause the condition or where the allele(s) have a higher prevalence in the general population than the condition itself.

“The... association data indicate that risk conferred by APOE differs between genders and possibly by ethnic groups”

-- Goldman et al. Genetic counseling and testing for Alzheimer disease: joint practice guidelines of the American College of Medical Genetics and the National Society of Genetic Counselors

“MTHFR polymorphism testing has minimal clinical utility and, therefore should not be ordered as a part of a routine evaluation for thrombophilia”

-- Hickey et al. ACMG Practice Guideline: lack of evidence for MTHFR polymorphism testing

Why is this recommendation important for patient care?

- Testing in these cases is not only unnecessary given the lack of clinical utility, especially for those conditions which are non-preventable but also because the results can cause unjustified anxiety
- There may be a role for genetic counsellors working to educate patients on the utility of genetic testing
- General practitioners could have a role in this process though one problem is that genetic testing services are nowadays ‘direct to consumer’