Tips for providers *before* ordering an Expanded Carrier Screening panel

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Before ordering EXPANDED CARRIER SCREENING, consider these **six** questions:

1) Have I taken an up-to-date three-generation family history?
   a. Would a referral to genetics be indicated regardless of expanded carrier screening results e.g. family history of a genetic condition, of an ethnicity where there is higher prevalence of certain conditions, history of a previous child with congenital anomalies?

For more on what red flags to target when taking a preconception family history, see the GEC-KO page on Reproductive Genetic Carrier Screening in Point of Care Tools.

2) Does the testing company also offer genetic counselling? This may be helpful to your patient if carrier screening is positive.
   a. Is this with or without an additional fee?
   b. Are the company's genetic counsellors board certified (designated as Certified Genetic Counselor (CGC) or Canadian Certified Genetic Counsellor (CCGC))?

3) Am I comfortable handling the results of this testing?
   a. Are variants of uncertain significance reported?
   b. Could I deal with unclear results?
   c. Genetics clinics may not be able to see your patient for the sole indication of a private genetic test result. Your patient should be made aware that access to formal genetic counselling may be limited.

4) Have I discussed the possibility of insurance discrimination with my patient?
   a. Carrier screening panels may diagnose an otherwise apparently healthy individual with an inherited condition e.g. hereditary hemochromatosis, familial hypercholesterolemia

5) Does my patient understand that:
   a. Screening is voluntary?
   b. A negative result does not eliminate the risk for a genetic condition?
   c. Accurate paternity is required for accurate reproductive risk assessment?

6) Do I and my patient have some familiarity with each disorder on the panel?
   a. Inheritance (e.g. recessive, X-linked)
   b. Onset (childhood vs. adulthood)
   c. Severity (treatment or management available, e.g. Tay Sachs disease vs. cystic fibrosis vs. factor V Leiden)

For further reading on expanded carrier screening, consider these articles:

Edwards JG, Feldman G, Goldberg J. et al. Expanded carrier screening in reproductive medicine-points to consider: a joint statement of the American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, National Society of Genetic Counselors, Perinatal Quality Foundation, and Society for Maternal-Fetal Medicine. Obstet Gynecol 2015; 125(3):653-62.

Henneman L, Borry P, Chokoshvili D, et al. Responsible implementation of expanded carrier screening. Eur J Hum Genet 2016; 24(6):e1-e12.

See the GEC-KO Education Module in Prenatal and Preconception Genetics http://geneticseducation.ca/education-modules/prenatal-genetics/ for more on expanded carrier screening education.

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