

Topics that may be included on the EBMG genetic counsellor science exam

1. Chromosomes.
 - a. Gross structure of chromosomes
 - b. Meiosis, including recombination
 - c. X-inactivation
 - d. Chromosomal abnormalities – translocations and aneuploidy.

2. Genes
 - a. Structure of genes
 - b. Types of mutations in single genes
 - c. General concepts – gene loci, allelic heterogeneity, locus heterogeneity
 - d. Positional and functional cloning.

3. Inheritance patterns
 - a. Mendelian inheritance patterns and assessment of risk based on these
 - b. Mitochondrial inheritance
 - c. Imprinting

4. Common laboratory tests
 - a. Karyotyping
 - b. FISH
 - c. PCR
 - d. Arrays
 - e. Sequencing.

This is an open book examination. You will be able to use textbooks or the Internet. Some questions may require the ability to use databases such as OMIM or Gene tests/Gene Reviews.