



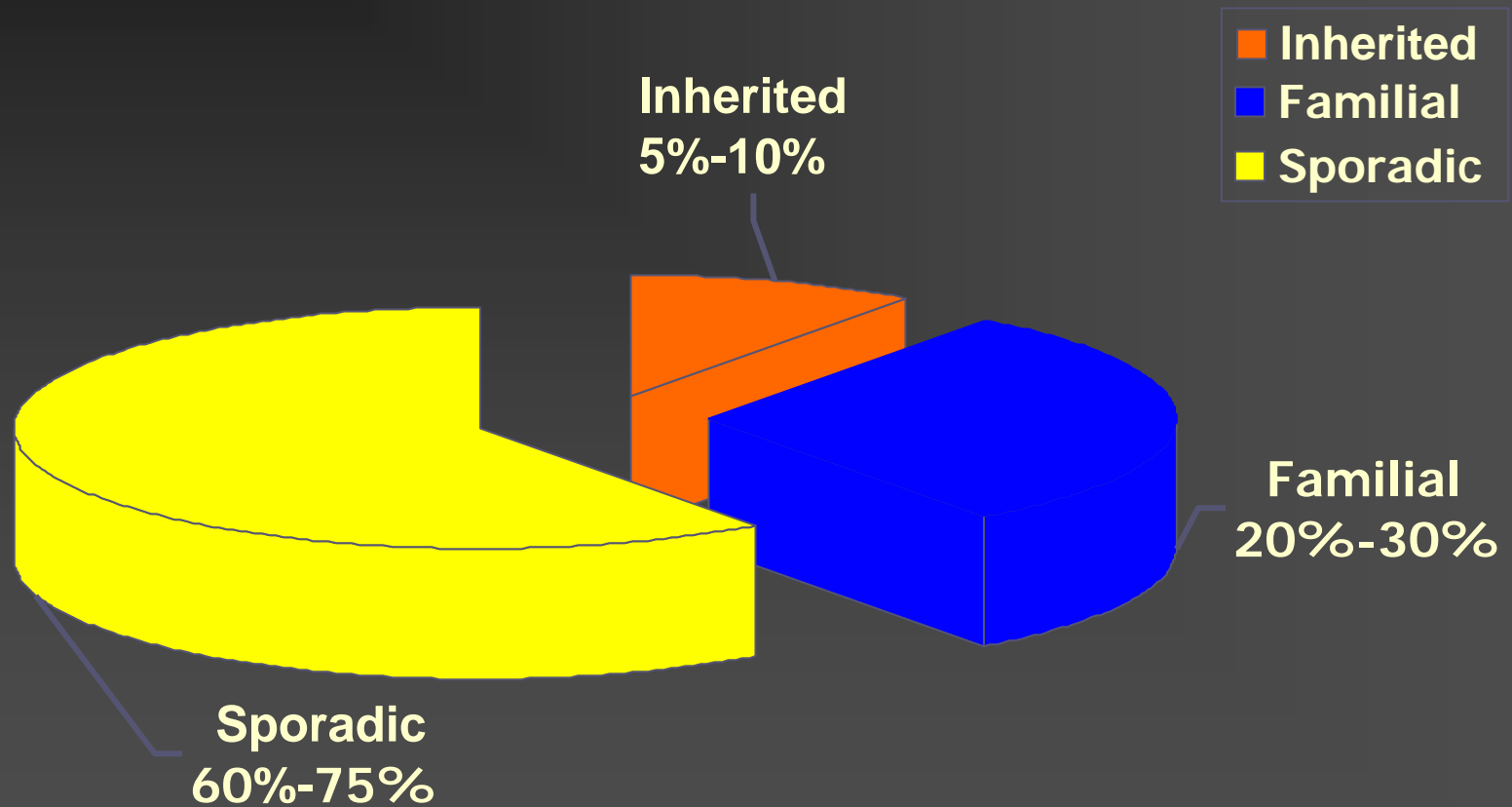
Beyond the Family History: Inherited Breast and Colon Syndromes

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Review of presentation

- Suspicious family histories
 - When/how to refer
 - Breast Cancer Syndromes
 - Colon Cancer Syndromes
 - FAP
 - HNPCC
 - Resources
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Most cancers are NOT inherited



Disease clustering

- Coincidental clustering of sporadic cases of a common condition
 - Due to a combination of genetic and lifestyle or environmental factors
 - Due to the presence of an inherited gene mutation
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How to identify if genetic risk?

- Pedigree intake
 - collect family history for three generations
 - relationships, vitals, health status (age onset)
 - both maternal and paternal sides
 - include unaffected relatives
 - document from more than one family member
 - confirm diagnoses
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When to refer for genetic consult

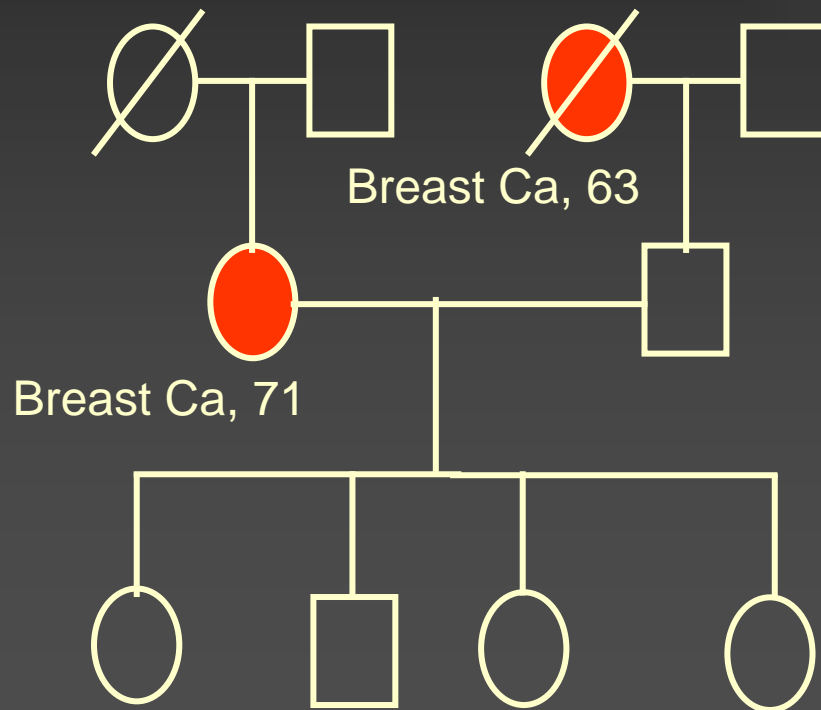
- From provider's perspective
 - Identification of suspicious pedigree
 - Unusual pathology report
 - From patient's perspective
 - Formally express concern
 - Informal comments
 - Anniversary dates
 - New diagnosis in family
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What is genetic counseling?

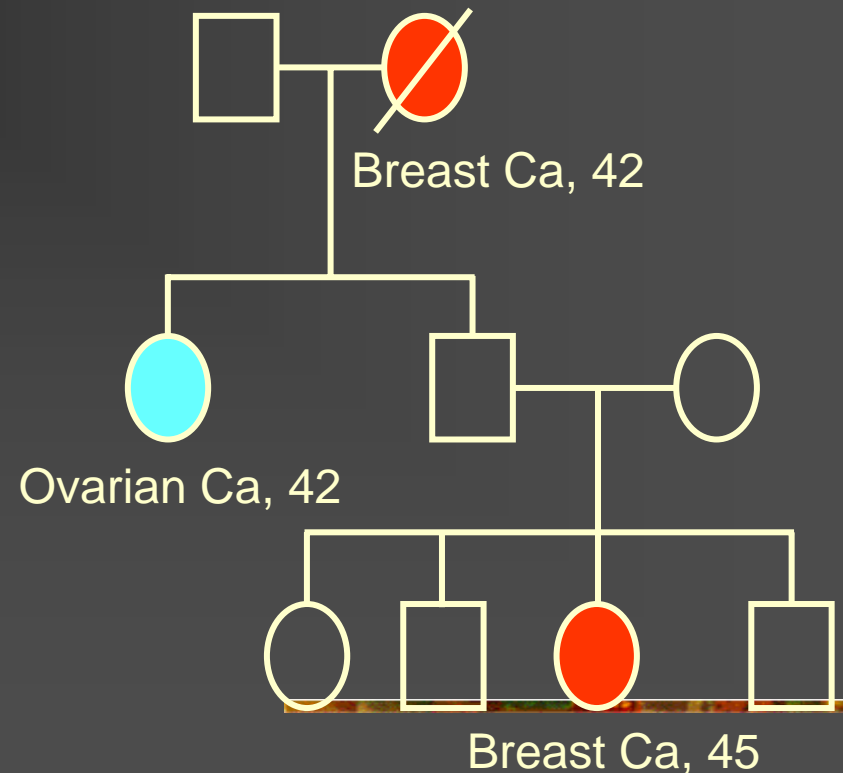
- Process of
 - Collecting family history, medical records
 - Assessing to establish risks
 - Evaluating psychosocial/family dynamic
 - Consulting
 - Counseling
 - Education – available from all care providers
 - Result - individual or family understands medical and scientific information about an inherited condition or birth defect – how to respond
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Sporadic and inherited cancer

Sporadic



Inherited



Breast risk numbers

- General population
 - 1 in 8 lifetime risk (12%)
 - First/second degree relative affected
 - two to three fold increased risk (20% - 30%)
 - Dominant mutation (*BRCA1*, *BRCA2*)
 - 60 to 80% lifetime risk
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Factors pointing to B1/2

- Multiple cases of early onset breast cancer
 - Ovarian cancer (with family history of breast or ovarian cancer)
 - Breast and ovarian cancer in the same woman
 - Bilateral breast cancer
 - Ashkenazi Jewish heritage
 - Male breast cancer
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Breast Ovarian Cancer Syndrome

- 1 in 800 in general population
 - Higher in Ashkenazi Jews (1 in 40)
 - Founder mutations in AJ
 - 187delAG, 5385insC in *BRCA1*
 - 6174delT in *BRCA2*
 - Other founder mutations
 - Dutch, Italian, Icelandic
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