Canadian Family Medicine Clinical Card

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Comprehensive Family History

Core Family History

Relevant health information from three generations including:
- grandparents, aunts, uncles, half-siblings, nieces and nephews
- cousins and great-grandparents, if available.

Key Elements[1,2]:

Personal Information:
- ❑ Names
- ❑ Ages
- ❑ Current health status
- ❑ If deceased: age and cause of death
- ❑ Ethnicity

Issues to explore:
- ❑ early onset of illness
- ❑ pregnancy Issues
- ❑ infertility
- ❑ miscarriages or still birth
- ❑ birth defects
- ❑ known familial diseases or conditions
  e.g. Cystic Fibrosis, Huntington Dz, familial ALS, Sickle Cell Dz)
- ❑ known nonmedical conditions
- ❑ consanguinity:

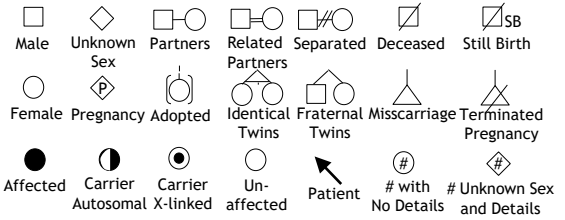
"Is there any chance that any of the couples in your family may be blood relatives?"

Red Flags

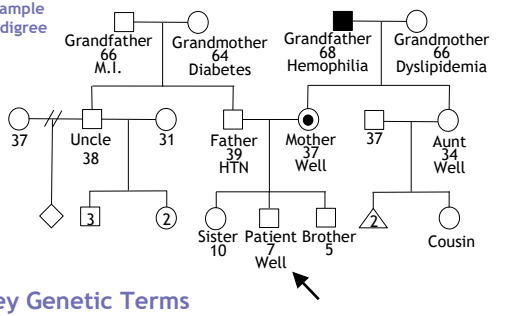
		FamHx significant for:	Dx To Think About:
General		two or more congenital anomalies +/- dev. delay or intellectual disability	genetic syndrome
General		multiple affected siblings or individuals in multiple generations	suggests genetic etiology
General		multiple child deaths, still born, miscarriages	suggests genetic etiology
General		extreme presentation of common conditions or pathology	suggests genetic etiology
Specific	Hereditary Cancers	early onset, colorectal cancer in multiple generations, usually in each generation	Familial Adenomatous Polyposis or Hereditary Non-Polyposis Colorectal Ca
Specific	Hereditary Cancers	early onset, >1 primary melanoma in patient, >1 family member with melanoma	Familial Melanoma
Specific	Hereditary Cancers	early onset, >1 primary breast ca in patient, >1 family members with breast ca, breast ca + ovarian ca, breast ca in males	Hereditary Breast and Ovarian Ca
Specific	Hereditary Cancers	≥2 endocrine neoplasias	Multiple Endocrine Neoplasia
Specific	Blood	recurrent, unusual, or early onset VTE	Factor V Leiden
Specific	Blood	significant bleeding history or sequelae	Hemophilia
Specific	Blood	anemia	Sickle Cell Dz or Thalassemia
Specific	Other	early onset (<65yrs) dementia	Early Onset Alzheimer Disease
Specific	Other	syncope, sudden cardiac death in family member, unexplained drowning, single car MVC	Heritable Arrhythmia/Cardiomyopathy

Constructing a Pedigree

Legend



Example Pedigree



Key Genetic Terms

Pattern	Features
Genotype	An individual's genetic makeup.
Phenotype	An individual's observed characteristics; based on genetics and environment.
Autosomal Dominant	Typically affects each generation. 50% likelihood of being affected.
Autosomal Recessive	Typically skips generations. 25% chance of being affected, 50% chance of being a carrier.
X-Linked Dominant	Females more likely affected. No affected sons of an affected male.
X-Linked Recessive	Males more likely affected. Can have affected males in each generation.
Mitochondrial	Affects males and females equally. Only passed on by mothers.
Expressivity	The phenotypic variability of a genetic disease.
Penetrance	The proportion of patients with a mutation that have a disease phenotype.

Key References: Genetic Alliance. (2010). Understanding Genetics: A District of Columbia Guide for Patients and Health Professionals. Washington, DC. *District of Columbia Department of Health*. Tarini B, McInerney J. Family history in primary care pediatrics. *Pediatrics*. 2013;132(Suppl3): S203-S210. Whelan AJ, et al. Genetic red flags: clues to thinking genetically in primary care practice. *Prim Care*. 2004;31(3):497-508.

GenetiKit. (n.d.) Family History Tool. Retrieved from www.mountsinai.on.ca/care/family-medicine-genetics-program/resources/FamilyHistoryTool.pdf. Genetics Home Reference. (2019). What are reduced penetrance and variable expressivity. Retrieved from https://ghr.nlm.nih.gov/primer/inheritance/penetranceexpressivity.