

## MEDICAL GENETICS ACADEMIC SESSIONS:

### Case Presentation:

Present a clinical case of a known or unknown genetic or metabolic disorder, which provokes interest from an ethical, clinical, diagnostic, genetic counselling, or psychosocial perspective. Topics to include:

- Presenting complaints and history
- Examination findings (if applicable)
- Differential diagnoses and investigations
- Genetic counselling issues
- Psychosocial issues
- Diagnostic testing issues
- Ethical Issues
- Summary

Title and objectives should be provided to the Chief Resident at least 1 week in advance

### Mini-Review:

Present a review on a genetic condition. Topics to include:

- History
- Prevalence, incidence, epidemiology – Include some/all of this information if it is particularly interesting or has a particular relevance to information you will be presenting at some point in your lecture
- Clinical features
- Natural history/prognosis
- Treatment/management/therapy
- Genetics (molecular, mechanism, recurrence risk)
- Pathophysiology (where relevant - embryology, protein structure and function)
- Diagnostic testing and other laboratory features (how it relates to disease process)
- Carrier/pre-symptomatic/prenatal and other diagnostic testing – include difficulties of these tests
- Current research – ex: what models are available and studied for the disease (findings), gene modification, gene therapy, drug trials,

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### Guideline Review:

Present a new guideline to review for a particular disease/clinical presentation. Recommendations are to be evaluated based on levels of evidence and compared /contrasted to current practice. Guidelines are usually identified and assigned through respective disciplines (ie: prenatal, metabolic, medical genetics, cancer), however a student may decide to present guidelines in consultation with discipline lead. The guideline and objectives should be provided to the Chief Resident at least 1 week in advance for distribution.

### Informal Case Rounds:

Participants should be prepared to present a short synopsis on interesting cases seen recently in hospital, in clinic, or in the laboratory in an informal, round-table format. Visual aids are not necessary. Possible cases could include those with important differential diagnoses, classic or variant presentations of known disorders, medical mysteries, or those with interesting social or ethical situations. The goal of this session is to keep the clinical genetics group abreast the interesting cases that are being seen in the various areas of our department and not to review specific syndromes.

**Journal Club:** See Guidelines for Journal Club