Medical Genetics in Undergraduate Medicine

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Section Leader without Separate Clerkship
Overall Goal of New Genetics Curriculum

• To integrate teaching of genetic principles, genetic diseases and genetic tests seamlessly into the new curriculum in all modules

• To create physicians who can utilize this knowledge of genetics in their clinical practice, regardless of their specialty
Scope of Genetics in Clinical Practice

• Informal survey of 2 journals from each of 6 specialties looking for articles relating to genetics in January, February and March, 2013 issues

• Total- 54 articles
  – Psychiatry- 8 articles
    • CJP- 4 articles
    • AJP- 4 articles
  – Internal medicine- 12 articles
    • JAMA- 8 articles
    • NEJM- 4 articles
Scope-2

- Pediatrics- 15 articles
  - J Ped- 11 articles
  - Pediatrics- 4 articles
- Anesthesiology- 5 articles
  - Anesthesia- 4 articles
  - Canadian J of Anes- 1 article
- Obstetrics and Gynecology- 7 articles
  - AJOG- 4 articles
  - Green journal- 3 articles
- Surgery- 7 articles
  - Annals of Surg- 4 articles
  - Br J Surg- 3 articles
Current Curriculum

• Genetics is primarily taught in:
  – Block 1: Structure/Function and Disease Mechanisms
    • 6 hours lectures
    • 2 hours tutorials
    • 1 hour assigned reading
  – Block 2: Human Development
    • 10 hours lectures
    • 6 hours tutorials
    • 2 hours assigned reading

• 1 or 2 hour lectures in the other blocks regarding a particular organ system ie musculoskeletal, respiratory, cardiovascular
New Curriculum

- M0 Foundation Module
  - 7 hours
- M1-7
  - Hours not yet assigned
New Curriculum Opportunities

- Relevant placement of genetics topics: students are able to give them appropriate attention.
- Scaffolded curriculum: complex topics and principles are revisited and clinical importance can be reinforced.
- Integrated curriculum: complex topics discussed in variety of contexts, reinforcing different principles like Consolidation, CP4, ELSI, Indigenous health.
- **Challenge**: delivery of concepts in an understandable and logical way without a dedicated course.
WRHA Program of Genetics and Metabolism

- Professional Appointments
  - Department of Pediatrics & Child Health
  - Department of Biochemistry & Medical Genetics
  - Diagnostic Services of Manitoba
- 5 clinical geneticists (MD)
- 3 laboratory geneticists (PhD)
  - Cytogenetics, Molecular DNA Diagnostic, Metabolic Laboratories
- 10 genetic counselors (MS or PhD)
Outline of Topics: Need to Know

• Structure, function and regulation of the genome-DNA, genes and chromosomes
  – Transcription, gene expression, RNA

• Natural genetic variation within populations and the unique populations in Manitoba
  – Types of mutation, polymorphisms, haplotypes, founder effects, consanguinity, genetic linkage
  – Population screening: prenatal, newborn, carrier, inborn errors of metabolism, presymptomatic

• Family history
  – pedigree drawing and analysis
Need to Know-2

• Traditional and nontraditional patterns of inheritance and their underlying mechanisms
  – Imprinting, mitochondrial inheritance, mosaicism, unstable DNA sequences & expansion, epigenetics

• Genetics in medicine
  – Genetic syndromes and birth defects, prenatal genetics, cancer genetics, metabolic genetics, genetic conditions in organ systems, EBM

• Laboratory genetics
  – Chromosome analysis, FISH, aCGH, SNP array, GWAS, exome sequencing, DNA tests, variant of unknown significance
Need to Know-3

• Genomic medicine
  – Personalized health care with focus on prevention
  – Pharmacogenetics
  – DTC tests- paternity, forensic, drug response, predisposition

• Ethics in genetics
  – Predictive testing, reproductive decision making, carrier testing of minors, genetic testing in research, DNA storage, stem cells, cloning
  – Cultural sensitivity/safety
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Examples of Content

• Consolidation
  – Interpretation of genetic lab reports (tutorial)
    • Variants of unknown significance
    • Predictive /presymptomatic testing
      – Oncology, adult onset disorders, prenatal testing
  • Screening tests

• Transition to Clerkship
  – Nondirective genetic counseling (assigned reading)
  – Giving bad news (tutorial, small groups)
  – Interviewing family with genetic disorder
Examples-2

- **Clinical reasoning**
  - CP₄ - differential diagnosis including genetic causes:
    - Cardiac murmurs, dementia, hearing loss, hirsutism, seizures, vision loss, hemoglobin disorders, developmental delay in children

- **Community health**
  - Epidemiology - genetic conditions in unique populations (tutorial)
    - Founder effect, cultural differences and sensitivity, screening

- **Clinical Skills**
  - Communicating bad news, taking family history
  - Self reflection about counseling styles
Examples -3

• **Professionalism**
  – ELSI- HCM with molecular confirmation done presymptomatically in children and telling them their diagnosis

• **Indigenous health**
  – Aboriginal health, healing and story telling and how it relates to genetics
  – Reproductive health, FASD

• **Scholarly Activity**
  – Many projects ongoing for student involvement
Examples-4

- **Surgery/Anesthesia**
  - Pharmacogenetics
  - Hereditary cancer family syndromes (HNPCC)
  - Congenital heart disease

- **Family Medicine/Psychiatry**
  - Child development
  - Prenatal genetics
  - Psychiatric presentation of genetic disorders
Examples-5

• Internal medicine/Emergency
  – Long QT, HCM, seizures, hypoglycemia,
  – Huntington disease -predictive testing
  – Hematology- thalassemias, sickle cell, hemophilias

• Obstetrics& Gynecology/Pediatrics
  – Prenatal genetics
  – Birth defects
  – Teratogens
  – Developmental delay
  – Dysmorphology