

Blended Learning Journey on Growth Disorders

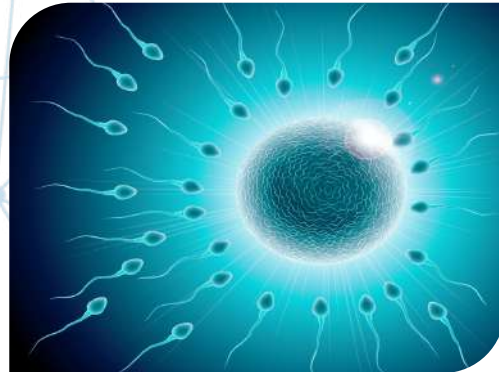


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Sperm and egg cells have methylated regions on their DNA

At fertilization, at loci where gene expression is restricted to either maternal or paternal genes, it is the methylation regions that denote which parental genes are expressed



Only having one active copy of genes means different proportions of “good” and “faulty” genes can be expressed in individuals with the same imprinting disorder

- There can be overlapping features with other ImpDs
- MLID (a subgroup of ImpD) can cause more than one ImpD in the same individual
- MLID can be associated with a history of maternal miscarriage
- **Diagnosis of ImpD can be challenging**

Imprinting Disorders: A Clinical Guide

13 imprinting disorders

Examples...

Prader-Willi syndrome
Angelman syndrome
Temple syndrome
Silver-Russell syndrome

Transient neonatal diabetes mellitus
Pseudohypoparathyroidism
Beckwith-Wiedemann syndrome

Causes profound effects on growth and metabolism, behaviours and neurocognition

ImpDs are often misdiagnosed, overlooked or delayed in diagnosis causing significant morbidity

Early diagnosis, initiating correct management and treatments all optimize outcomes

