

## **The cardiac manifestations of the inborn errors of metabolism**

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### **Abstract:**

The inborn errors of metabolism (IEM) exhibit protean clinical manifestations. Cardiac disease either in isolation or as part of a wider multisystem disease process, are frequently observed in the IEM. Cardiac presentations associated with the IEM include, arrhythmias, dilated cardiomyopathy, hypertrophic cardiomyopathy, non-compaction, valvular disease, endocardial fibroelastosis, and aortic root pathology. IEM groups commonly associated with cardiac disease include the mitochondrial respiratory chain defects, fatty acid oxidation defects, glycogen storage diseases, lysosomal storage diseases, defects of organic acid and amino acid metabolism, and defects of post-translational modification. This presentation will provide an update of the IEM disease process associated with cardiac disease, outlining a simple clinical approach to delineating an overarching diagnosis; especially in disease processes where a rapid diagnosis is essential, e.g., Pompe Disease.

### **Biography**

David Coman is a Paediatrician, Metabolic Physician and a Clinical Geneticist working in clinical practise in Brisbane Australia. He has a public appointment at the Royal Children's Hospital in Brisbane, and is the Medical Director of Paediatrics at the Wesley Private Hospital in Brisbane. He is interested in teaching and is the Academic lead for Paediatrics in the Uniting Care Clinical School, Brisbane. He has a particular research interest in disorders associated with aberrant post-translational modification, epigenetics modification of single gene disorders, and novel disease discovery. He has active interests in rare disease advocacy, and runs philanthropic general paediatric clinics to the Western provinces of the Solomon Islands.

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