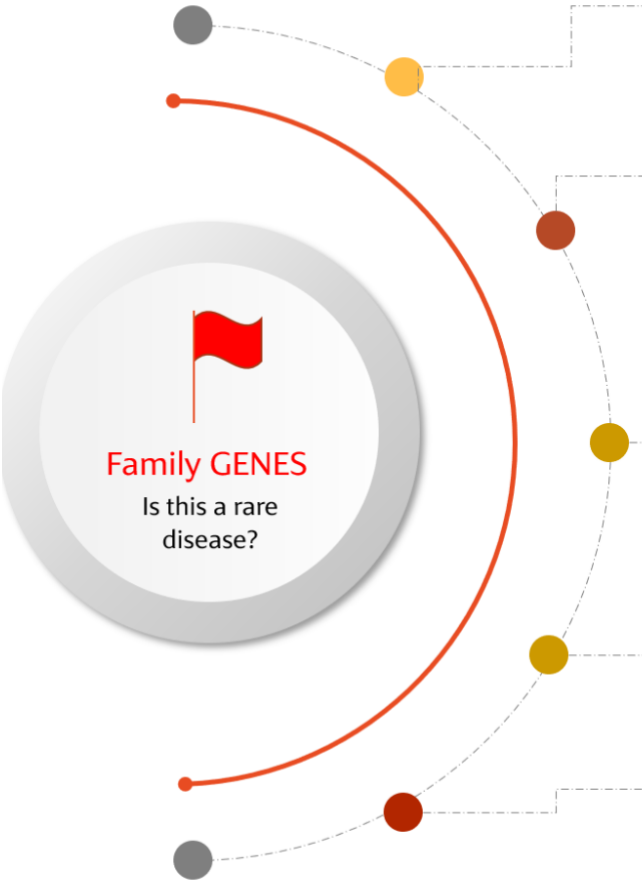
RArEST

Rare Disease Awareness, Education, Support, and Training

Think Rare by thinking Family GENES



G — **Group of congenital anomalies**
Common anatomic variations are, well, common; but two or more anomalies are much more likely to indicate the presence of a syndrome with genetic implications

E — **Extreme or exceptional presentation of common conditions**
Early onset cardiovascular disease, cancer, or renal failure. Unusually severe reaction to infectious or metabolic stress. Recurrent miscarriage. Bilateral primary cancers in paired organs, multiple primary cancers of different tissues

N — **Neurodevelopmental delay or degeneration**
Developmental delay in the paediatric age group carries a very high risk for genetic disorders. Developmental regression in children or early onset dementia in adults should similarly raise suspicion for genetic etiologies

E — **Extreme or exceptional pathology**
Unusual tissue histology, such as pheochromoctyoma, acoustic neuroma, medullary thyroid cancer, multiple colon polyps, plexiform neurofibromas, multiple exostoses, most paediatric malignancies

S — **Surprising laboratory values**
Markedly abnormal pathology results

Family history
Multiple affected siblings or individuals in multiple generations.
Remember that lack of a family history does NOT rule out genetic causes

Baynam, G. (2015). A Diagnostic Odyssey – Red Flags in the Red Sand. *Australian Medical Association (WA).* *Amendment made by Dr Baynam 5th December 2016.

Evans, W., McKay, L., Qureshi, N., 2021. Identifying, managing, and supporting patients with a rare disease. *InnovAiT: Education and inspiration for general practice*, 14, 92–99. doi:10.1177/1755738020974411