Case Presentation
Associate Professor
Norsarwany Mohamad
USM
• Introduction

• Not uncommon case, often not fully investigated

• Message: require communication and discussion among managing team
2 yrs and 1 month NH, referred case from HKL at the age of 11 months

- Admitted HKL at 10 months old for bronchopneumonia, noted pallor with hepatosplenomegaly (liver 2cm, spleen 1cm)
- Hb 7.4 g/dL, MCV 44.1 fl, and MCH 13.6 pg
- Se Ferritin 186.1 μg/L, Serum Iron 9.3 μmol/L, TIBC 44 μmol/L
- Hb analysis suggestive of Hb E with another possible Hb variant
- Father HbE trait, mother Hb constant spring trait
• 2 elder sisters with HbE trait
• Patient never been transfused
• Clinically active, weight < 3<sup>rd</sup> centile, height at 10<sup>th</sup> centile
• Now liver just palpable, spleen not palpable
• The double heterozygous state of α/β thalassaemia may alter the haematological indices and modify the phenotype.
• Hence characterization of the molecular defects is essential
• Prenatal diagnosis and genetic counselling
• South East Asia: recommend testing for both
• Cap+1 (A-C) is a silent mutation
• Produces very less clinical severity and Hb A₂ levels are also remain in normal range
• Individuals with Cap+1 mutation may produce beta-thalassemia intermedia if co-inherit with other beta mutation
• Malaysian study (Chong et al 2006): 3.5% of β-thalassaemia have concurrent carriers of α-thal with SEA deletion

• Wee et al (2008): α-thal was found in 12.7% β-thalassaemia carrier (\(\alpha^{3.7}\) & \(-\alpha^{4.2}\) deletions, Hb Constant Spring)

• In our case: **Compound heterozygous E β-thalassaemia co-inheritance with α-thalassaemia-SEA deletion**
• Ref:

• Alpha-Thalassaemia in Association with Beta-Thalassaemia Patients in Malaysia: A Study on the Co-Inheritance of Both Disorders (wee et al, 2008)

• Screening of concurrent α–thalassaemia in β thalassaemia carriers (Chong et al, 2006)