



Improvement in Prenatal Screening, Diagnosis & Management of α Thalassemia



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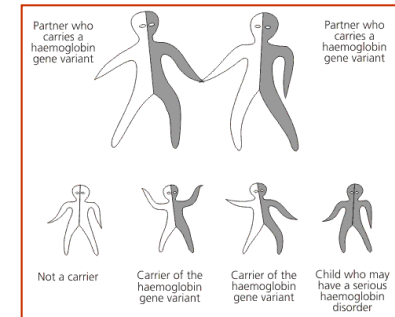
Maternal Fetal Medicine Team



PATHOLOGY

Prevention of Hb Bart's Disease

- α thal is common (4.5%)
- If α thal couples, 25% of their fetus will be affected.
- An affected baby with severe anemia will die in utero or shortly after birth.
- Mother may suffer from severe hypertension, bleeding or rarely death.





Screening

Past

No DNA analysis for suspected α thal in QEH, need send samples to TYH lab

Initiative (1)

- DNA analysis by Patho Dept QEH unless invasive test or inconclusive result
- 2011-13, of 18,623 screened pregnant women, 13.3% had low mean corpuscular volume, and 5.5% had α thal.
- **Sending out samples reduced by 76.6%**

Diagnosis

Past

For α thal couple, invasive prenatal testing was the only option with risk of miscarriage

Initiative (2)

- USG exclusion of Hb Bart's disease, and avoid invasive test if normal findings



- Of 57 α thal couples, 44 monitored by USG alone.
- Invasive test was avoided by 77.2% without missed cases at birth.

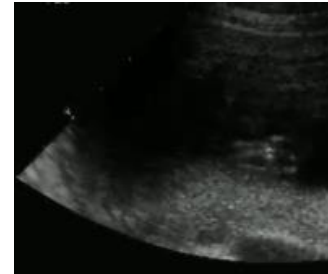
Management

Past

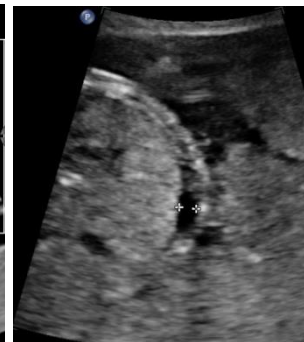
Termination is the only option for an fetal Hb Bart's disease

Initiative (3)

- In utero transfusion (IUT) is an alternative after counseling.



- Of 10 Hb Bart's disease, **one treated** by IUT with Patho and blood bank.



Pre- IUT

Post- IUT

One year

Detection of Two Unusual Hb Bart's Hydrops



- Partners' MCV were normal
- One due to uniparental disomy
- The other due to non-paternity
- Importance of routine scan to pick up hydrops

Kou KO, Lee H, Lau B, Wong WS, Kan A, Tang M, Lau ET, Poon CF, Leung KY
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Conclusion



- With collaborative efforts, we have improved the prenatal screening, diagnosis and management of α thal by improving workflow, reducing unnecessary invasive testing, and performing in utero transfusion.



Thank You

