Dysfibrinogenemia –
an unusual case.

By Katarzyna Mayger
Acknowledgement-
Dr Bella Madan
History

- A 33-year old woman who is a harpist in her second pregnancy.
- Diagnosed with dysfibrinogenemia during bleeding from a bout of gastroenteritis in 2009, with Clauss fibrinogen of 0.96g/L.
- Mild bleeding history with menorrhagia, epistaxis, easy bruising.
- Genetic defect: AαARG16.
- Previously treated with fibrinogen concentrates with an adequate response.
- Sub-optimal response to 4g of fibrinogen in previous pregnancy during labour in 2012. Denied epidural for pain relief.
Laboratory data-pharmacokinetic study during third trimester

Baseline (pre-infusion)
- PT Ratio: 1.3 (RR: 0.8-1.2)
- APTT Ratio: 1.0 (RR: 0.8-1.2)
- Clauss Fib: 0.6 (RR: 1.7-3.9) g/L
- Fib Ag: 4.36 (RR: 1.6-4.0) g/L
- Platelet count: 179x10^9 (Normal)

Post-infusion with 4g of fibrinogen
- Clauss Fib: 0.8 g/L at 20min, 1hr, 2hr, 3hr, 4hr and 5hr
Mixing studies 1:1 with Standard Human Plasma

- Pre-dose: Expected Clauss Fib: 1.85 g/L
  Measured Clauss Fib: 1.01 g/L.

- Post-dose: Expected Clauss Fib: 2.05 g/L
  Measured Clauss Fib: 1.26 g/L.

- Immediate and incubated Clauss fibrinogen mixing studies on pre-treatment and 20 min post-treatment samples suggest inhibitor.

- In view of patient's appreciable baseline fibrinogen activity, quantification in a Bethesda-type assay not possible.
Delivery and follow up

- Delivery via Caesarean section due to unstable lie.
- Tranexamic acid given 1g QDS starting 7 days before the procedure.
- No haemorrhagic complications.
- Neonate has low fibrinogen – further tests pending.
- Follow up visit scheduled to repeat pharmacokinetic studies on mother.