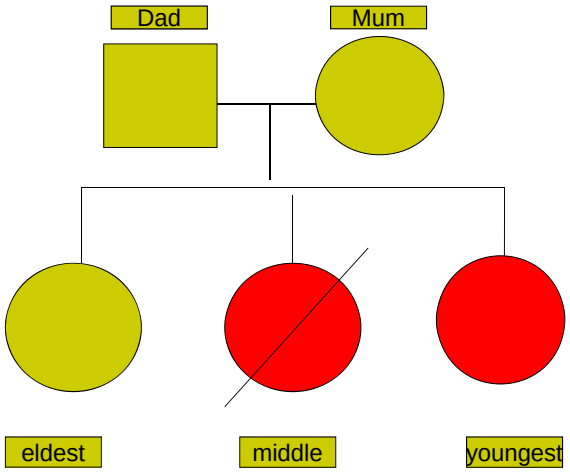
Glanzmann thrombasthenia

Rarer Bleeding disorders
HFA conference
Penny McCarthy
October 2011

Glanzmann thrombasthenia

- Due to platelet membrane GpIIb/IIIa resulting in
  - a platelet aggregation defect
  - No platelet plug formation at site vascular injury
  - Platelet count is normal
- Characterised by
  - Excessive menstrual blood loss
  - Bleeding from mucous membrane
  - Major haemorrhage following trauma or surgery

- First identified by Dr. Glanzmann in children from a Swiss Village
- More common where marriage between blood relatives is common
- Carried on chromosome 17, effects men and women equally
- Autosomal recessive
- Incidence is 1:1,000,000



Youngest

- Tested at birth as sister had previous diagnosis
- No serious problems in childhood
- Blood transfusion and platelets at menses managed with hormone therapy
- Transferred Nov 99 at 21yo with sister who required surgery
- 2004 sister died following BMT
- 2005 R/O of umbilical endometrioma
- 2005 -2007 moved to UK

Report from U.K

- Ceased pill, Mirena due to hormones SE
- 2005 urinary retention abdominal pain
- Ruptured corpus luteum
  - Rx TA, blood, HLA matched platelets
  - Mirena removed due to ongoing SE
  - Commenced pill and Zoledex

2007 Pregnancy???

Questions

- How to become pregnant
- Safety of foetus
- Safe delivery

- contacted Dr. Manchu Poon International Glanzmanns registry and Dr Yves Laurian in France
- Risks
  - Platelet antibodies
  - Foetal death or ICH
  - Maternal death from uncontrolled bleeding

IVF

- IVF specialist consulted
- Combined meeting
  - Youngest and her husband
  - IVF specialist
  - Anaesthetist
  - Haematologist

Risk of bleeding so great that pregnancy was not an option

What now??

- Remain childless
- Fostering
- Adoption
- Surrogacy

Australia, Russia, India

Happy Ending!