Mild Haemophilia

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Presentation of mild Haemophilia

- Testing because of the family history
- Bleeding following surgery or dental extraction
- Post traumatic bleeding
- Bruising
- Mucosal bleeding
- Menorrhagia

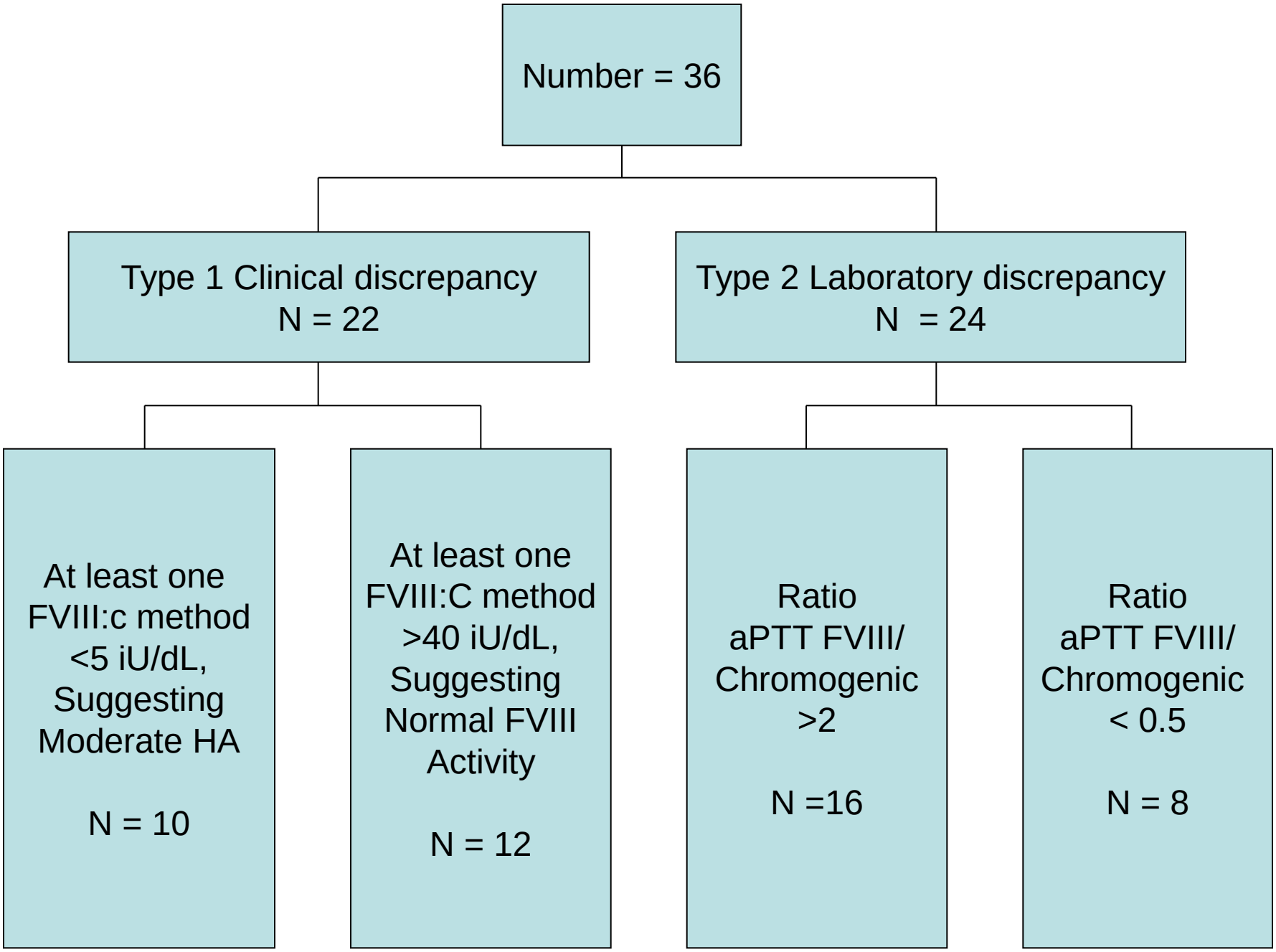
Mild Haemophilia patients at PMH

- 26 severe Haemophilia A (factor VIII <1%)
- 7 moderate Haemophilia A (2% – 5%)
- 21 mild Haemophilia A (5% - 40%)
- 2 type 2N von Willebrand's disease
- 2 severe Haemophilia B (factor IX <1%)
- 2 moderate Haemophilia B (2% - 5%)
- 3 mild Haemophilia B (5% - 40%)

Discrepancy with factor VIII by Chromogenic assay

- Factor VIII 6%
- Factor VIII 23%
- Factor VIII 12%
- Factor VIII 25%
- Factor VIII 9%
- Factor VIII 23%
- Factor VIII 28%

- Chromogenic 2%
- Chromogenic 16%
- Chromogenic 5%
- Chromogenic 70%
- Chromogenic 6%
- Chromogenic 7%
- Chromogenic 8%


Number = 36
Type 1 Clinical discrepancy
N = 22
Type 2 Laboratory discrepancy
N = 24
At least one FVIII:c method <5 iU/dL, Suggesting Moderate HA
N = 10
At least one FVIII:C method >40 iU/dL, Suggesting Normal FVIII Activity
N = 12
Ratio aPTT FVIII/ Chromogenic >2
N =16
Ratio aPTT FVIII/ Chromogenic < 0.5
N = 8

Twins with mild Haemophilia A

- Identical twins - diagnosis because of the family history. Elder brother effected.
- No previous family history of Haemophilia
- Factor VIII levels 15% (similar result by chromogenic assay).

  mutation c.329T>G (p.Met110Arg).

  DDAVP response 18% - 36%

Progress Twin 1.

- Multiple epistaxis – tried Tranexamic acid, nasal cautery, DDAVP, remained a clinical problem with school absences etc.
- Commenced r FVIII (Xyntha) given weekly. Epistaxis improved.
- Presented with ankle pain. No evidence of haemarthrosis.
- Dental extraction caused unexpected bleeding.

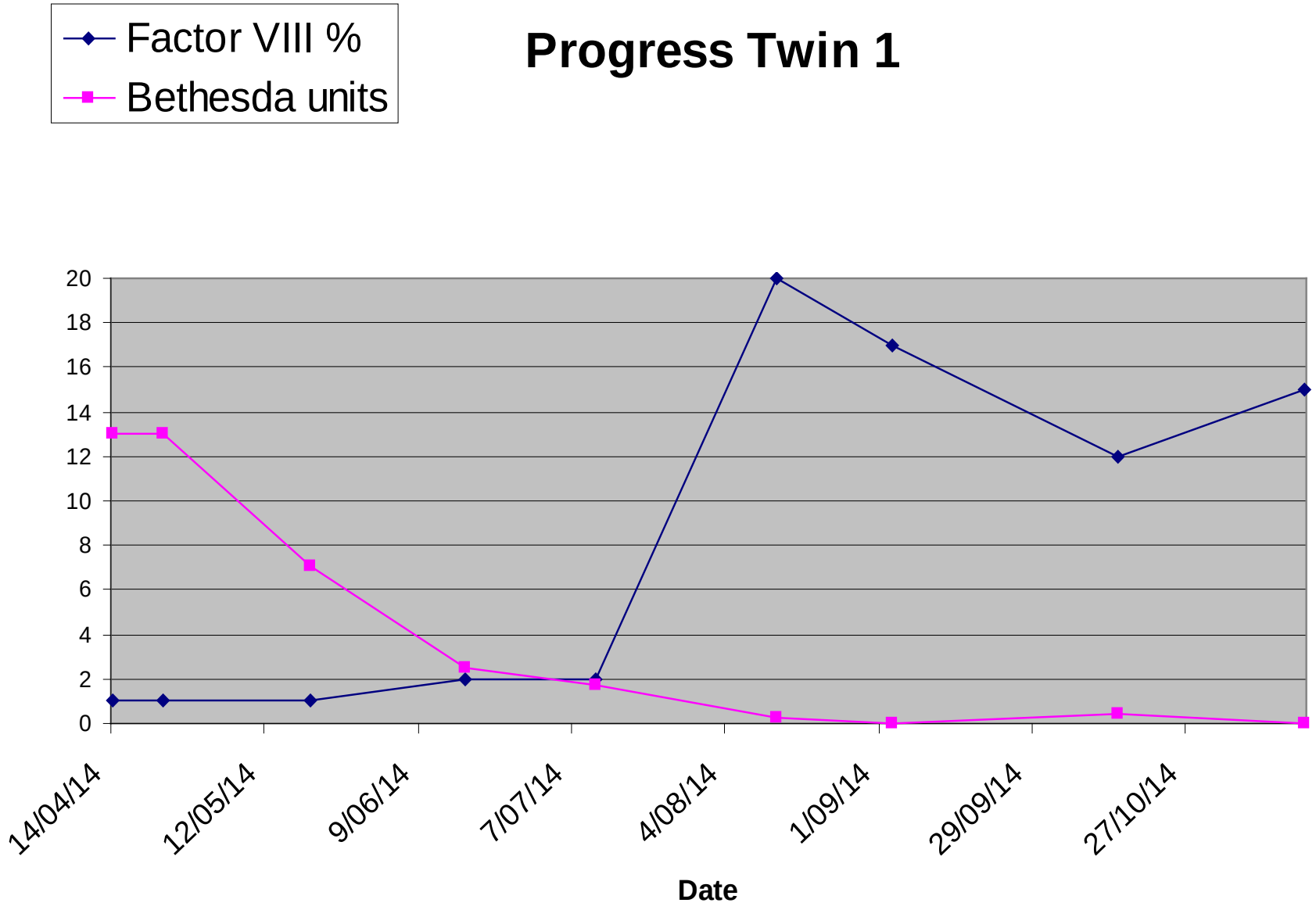
Progress Twin 1.

- Found to have an inhibitor >10 Bethesda units with factor VIII level now 1%.
- Given r FVIIa with control of mouth bleeding.
- Decision to commence ITI with r FVIII 100iU/Kg by peripheral vein.
- PICC line inserted.
- Broviac catheter inserted.

Progress Twin 1.

- Broviac catheter removed due to infection.
- Port inserted.
- ITI continued, bleeds controlled with r VIIa.
- ITI ceased after 6 months, prophylaxis continues at 50iU/Kg 2x/week.

Progress Twin 1



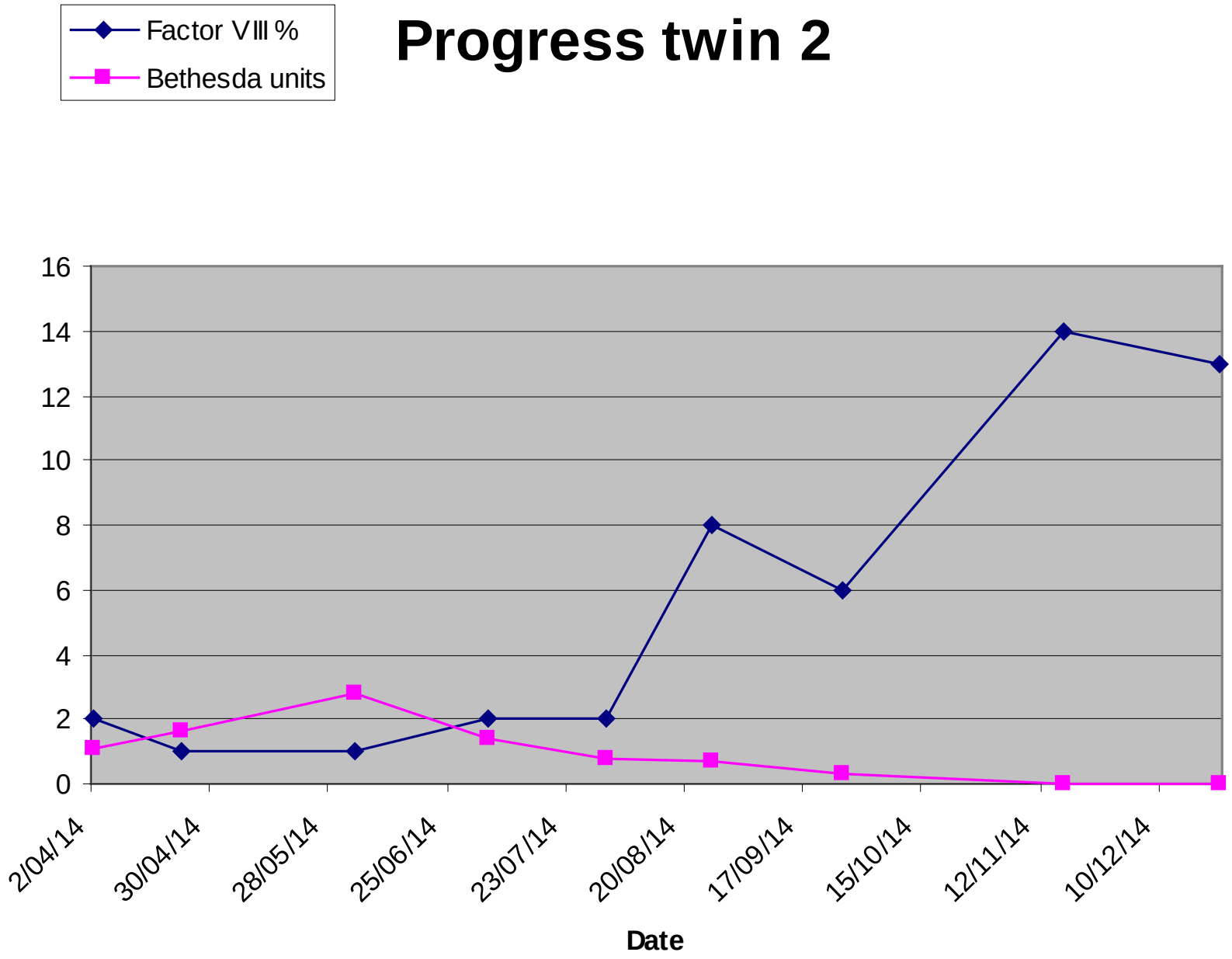
Progress Twin 2.

- First exposure to r FVIII aged 2 for head injury.
- DDAVP used for dental surgery.
- Recurrent epistaxis, tried Tranexamic Acid, nasal cautery, DDAVP.
- Commenced on r FVIII weekly with improvement in the epistaxis.
- Brother found to have an inhibitor.

Progress Twin 2.

- Inhibitor level found to be 2.2 Bethesda units with factor VIII 1%.
- Decision to treat with r VIIa and withhold further factor VIII.
- 2 hospital admissions for muscle bleeds treated with r VIIa.
- Forearm bleed treated with r VIIa.
- Remains off r FVIII.

Progress twin 2



ITI v No treatment

- ITI need for Port insertion. Hospitalisation for infections associated with Port.
- No difference in bleeding rate.
- Similar rate in fall of the inhibitor level.
- Future tendency for inhibitor development unknown.