Afibrinogenaemia

A case study from a nursing perspective.

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Overview

- What is Afibrinogenaemia?
- How rare is it?
- What are some of the issues for patients?
- What role do nurses play?

Afibrinogenaemia

- Autosomal recessive bleeding disorder.
- Lack of fibrinogen (Factor I)
- Occasional spontaneous muscle bleed but most often excessive prolonged bleeding and bruising after trauma.

Which Factor?

Table 1: Characteristics of Rare Clotting Factor Deficiencies

MISSING FACTOR	INCIDENCE*	INHERITANCE	SEVERITY OF BLEEDING	TREATMENT
Factor I Afibrogenemia Hypofibrogenemia Dysfibrogenemia	5 in 10 million not available 1 in 1 million	Autosomal recessive Recessive or dominant Recessive or dominant	Usually mild, except in afibrinogenemia	• Fibrinogen concentrate • Cryoprecipitate • Fresh frozen plasma
Factor II	1 in 2 million	Autosomal recessive **	Usually mild	• Prothrombin complex concentrate • Fresh frozen plasma
Factor V	1 in 1 million	Autosomal recessive	Usually mild	• Fresh frozen plasma
Combined factor V and factor VIII	1 in 1 million†	Autosomal recessive‡	Usually mild	• Fresh frozen plasma • Factor VIII concentrate • Desmopressin
Factor VII	1 in 500,000	Autosomal recessive **	Severe when factor levels are low	• Recombinant factor VIIa concentrate • Factor VII concentrate • Prothrombin complex concentrate • Fresh frozen plasma
Factor X	1 in 1 million	Autosomal recessive	Moderate to severe when factor levels are low	• Prothrombin complex concentrate • Fresh frozen plasma
Combined deficiency of vitamin K-dependent clotting factors	not available	Autosomal recessive **	Usually mild, but a few families have reported very low levels and more severe symptoms	• Vitamin K • Prothrombin complex concentrate • Fresh frozen plasma
Factor XI	1 in 100,000	Recessive or dominant	Mild to moderate when factor levels are low	• Factor XI concentrate • Antifibrinolytic drugs • Fibrin glue • Fresh frozen plasma
Factor XIII	1 in 3 million	Autosomal recessive	Severe	• Factor XIII concentrate • Cryoprecipitate • Fresh frozen plasma

* Estimates only
** Can also be acquired later in life because of another medical condition, certain medications, etc.
† 1 in 100,000 in some populations, including Israel, Iran, and Italy
‡ Very rarely, factor VII deficiency can be inherited separately from only one parent

How rare is rare?

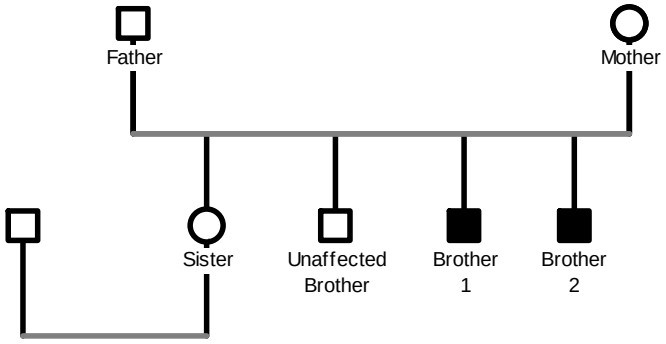
- Australian population June 2010:
  22,500,000
- Registered patients with Afibrinogenaemia June 2010:
  5

That makes them 1 in 4.5 million!

Case study

- 2 Brothers – aged mid twenties.
- Born and raised in Australia by immigrant parents of Spanish and Argentinean background.
- One older brother – not affected
- One older sister – normal fibrinogen levels but possible carrier (current tests being done to isolate gene mutation).

Family Tree



How was it diagnosed?

- Brother 1 presented to Haematologist at Children's Hospital in Brisbane at age 3 days with prolonged bleeding from heel prick site.
- Also bled from umbilical vein where coag tests subsequently taken.
- "Lab tests revealed a complete absence of fibrinogen."
- Brother 2 born two years later with same presenting signs.
- No known family history

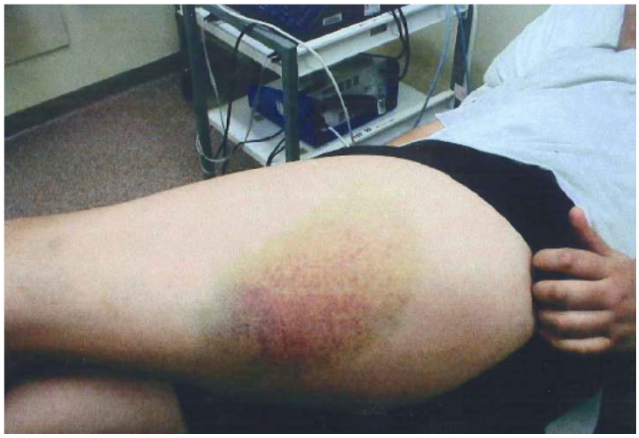
The start of an eventful journey..

- Mum and Dad struggled with the diagnosis in the beginning.
- Through necessity and with the help of the Haematologist at RCH, they developed a routine for hospital visits.
- Consistent visits approx every 6 to 8 weeks throughout lives.
- Active boys keen on sports.

Growing up with Afibrinogenaemia: Nursing Entries in Case notes.

- Fell out of pram while being pushed by sister
- Fell from shopping trolley yesterday
- Kicked in the calf at soccer
- Hit in face playing basketball
- Hit on left hand while playing hockey
- Fell when playing with brother – twisted ankle
- Pulled own tooth out last night – has not stopped bleeding.
- Cut foot on a rock while climbing
- Hit right knee on bike handlebar

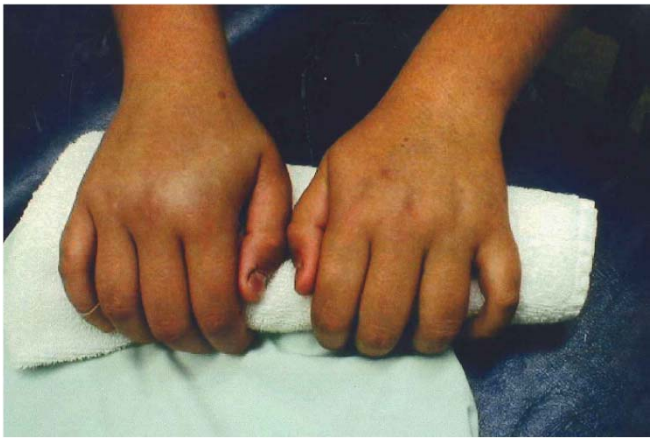
Sporting injury…



And it goes on……

- Hit right hand on a metal pole whilst swinging it around
- Bruising after go-kart racing
- Laceration (glass) to side of left foot
- Hit to R) side of face. Bruising to ear.
- Assault – nasal fracture, injury to one tooth and stitches to fingers of left hand.
- Dropped barbell on self at gym.
- Old shin injury but happens to be going racing in V8 cars tomorrow.

Hazards of play…



Treatment

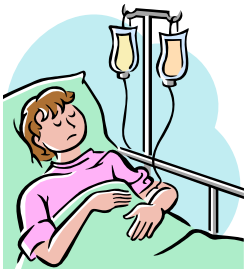
- Rest, Ice, Compression, Elevation
- Cryoprecipitate
- Physiotherapy
- Fibrinogen concentrate

Treatment - Cryoprecipitate

About Cryoprecipitate

Blood product from donors

- 1-2 hours outpatient time
- Takes 20 mins to thaw from frozen
- 15 bags per treatment
- Need IV cannula
- Allergic reaction
- Hydrocortisone and Phenergan pre-meds.
- Drowsiness – no driving

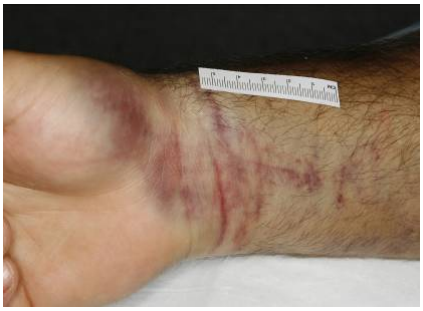


Fibrinogen Concentrate

- Fibrinogen concentrates are available in some parts of the world.
- Derived from pooled plasma.
- This product is unfunded by the National Blood Authority in Australia at present.

What are the boys up to today?

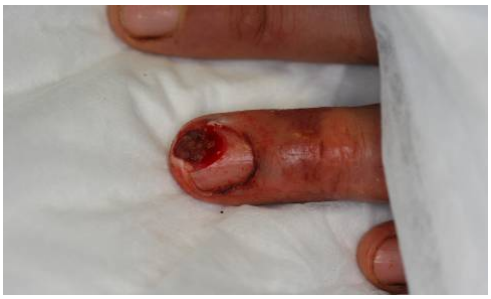
- Brother 1 works with older brother in **panel beater** workshop



- **Recent Nursing Note**: Dropped car bonnet on both hands

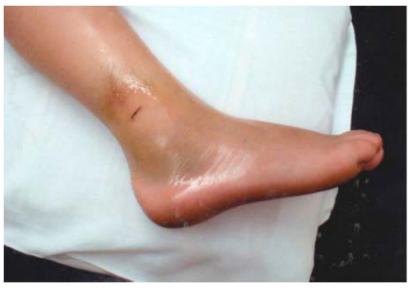
Occupational Hazards!

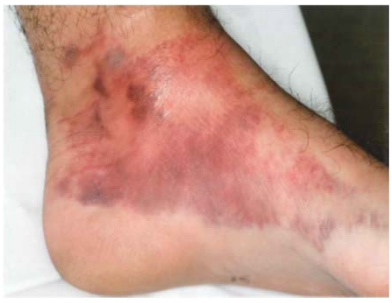
- Brother 2 works as **head chef** at inner-city pub.



- **Recent Nursing Note**: “Chopped finger tip while cutting capsicum”.

What has changed?





What has changed?

- The causes of the injuries!
- Funded treatment for Afibrinogenaemia has not changed in Australia. Cryoprecipitate is still the treatment available in Australia.

Points to take away

- People with bleeding disorders can encounter complications when accessing health and medical care, often having to explain things to health professionals.
- This is particularly the case for people with rare disorders such as afibrinogenaemia.
- Brother 2 is keen to move interstate, he is considering his options.

Continued…

It is important that nurses help **teach** and **guide** patients:

- To have a good understanding of their own disease and treatment. This helps them advocate for themselves.
- Always to put safety first (We are working on this!)

Thank you

- References:

- Mannucci, P. Duga, S and Peyvandi, F (2004) Recessively inherited coagulation disorders. Blood 104: 1243-1252.
- Kroll, M (2001) Manual of Coagulation Disorders, Blackwell Science, Massachusetts.
- Peyvandi, F. Cattaneo, M. Inbal, A. De Moerlooses, P & spreafico, M (2008) Rare Bleeding Disorders. Haemophilia 14:3, 202-210.

- Questions?