Case Report
Inhibitor Against Factor VII

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Factor VII

- Vitamin K-dependent glycoprotein
- Chromosome 13 at 13q34
- More than 130 mutations
- The most common RBD
- Prevalence of 1:300,000 – 1:500,000
- Autosomal recessive
- Poor correlation between FVII levels and bleedings
- Epistaxis, menorrhagia and other mucosal bleeding
Diagnosis & Treatment

• Isolated prolongation of the PT
• Other tests are usually normal
• FVII levels are low at birth
• It is important to exclude vitamin K deficiency
• Substitution therapy is the main therapeutic option
• Tranexamic acid in asymptomatic patients
• Plasma FVII half-life is short (4 hr)
• rFVIIa is the treatment of choice
• Fresh Frozen Plasma (FFP) had been widely used in the past
Case 1

- 4 year old boy
- Presented with hydrocephalus during infancy
- Down syndrome
- When he was 2 years old he developed F VII inhibitor with level of 191 BU
- Finally he died of severe intra-abdominal bleeding and serious sepsis in a general hospital while receiving high dose of rVIIa and FFP.
Case 2

- 8 year old boy
- presented with intracranial hematoma, hydrocephalus and melena at the age of two weeks
- The intracranial shunt placement was done after establishing the diagnosis under coverage of fresh frozen plasma (FFP) to correct his prolonged PT
- The patient was diagnosed as a congenital FVII D at the age of 40 days.
- Patient had intermittently received rVIIa, 1mg twice a week to prevent further bleedings
Case 2

- In March 2012, he came to emergency ward with chief complaint of **hemarthrosis and chronic synovitis** and underwent an arthroscopy on his right knee in a general hospital.

- In June 2012, patient was referred to another hospital with chief complaint of epistaxis and right knee hemarthrosis and he received weekly chemical (Rifampicine) synovectomy under coverage of 2mg rVIIa for several weeks.

- In August 2012, patient developed inhibitor to FVII, 170 BU.

- We treated him with **aPCC (Feiba)** and short term of oral **corticosteroid** therapy along with physiotherapy in our center.

- Two months later, level of inhibitor reduced to less than 20 Bethesda units and until **now he has treated with Feiba, on demand** and his musculoskeletal bleedings stopped.
Genetics

• Genomic DNA was extracted and a multiplex conformation sensitive gel electrophoresis (CSGE) showed abnormal profile.

• Genetic analysis has been performed by direct sequencing on an automated sequencer (ABI 3130 DNA analyzer, Applied Bio systems, Foster City, CA, USA)

• We have identified a c.9711 deletion C in a homozygous state in the exon 7 of F VII gene in both patients.
Take home message

• Start *prophylaxis* as soon as possible

• If you find changes in bleeding pattern (Hemarthrosis) think about INHIBITOR !?

• If you find this mutation (*c.9711 del C in exon 7*), probably is better to **postpone the prophylaxis**.
Thank you for your attention!