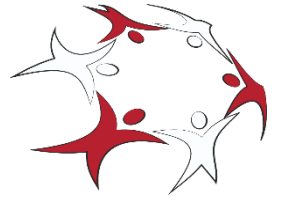


Genotyping and carriers

World Haemophilia Day
April 19th 2017

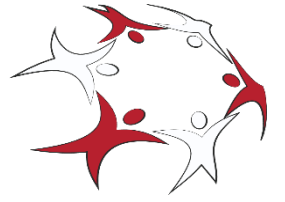
Naja Skouw-Rasmussen
EHC Steering Committee
Member

Recommendation 6



- **Genotype analysis** should be offered to **all patients with severe haemophilia**.
- **Genetic counselling to women** in the wider kindred.
- Genotyping desirable in cases of **mild** and **moderate** haemophilia and for patients with **other congenital bleeding disorders**.

Genotype analysis – Why?

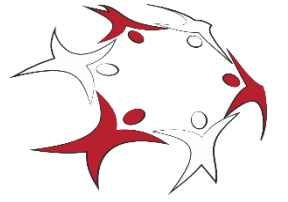


- Association between genotype and phenotype
- Individualise treatment
- Risk of inhibitor
- Family planning

Genetic counselling to women - Why?



- Genetic testing is the only test that provides certainty for being a carrier without bleeding episodes.
- In terms of reproduction:
 - Knowledge of risks about if the child will have a bleeding disorder.
 - Informed decision making about potential termination.
 - To prepare for child birth.

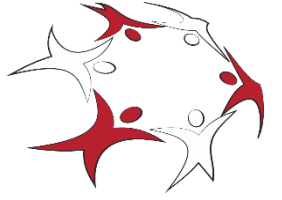


Carrier

Mild and moderate haemophilia
Other congenital bleeding disorders



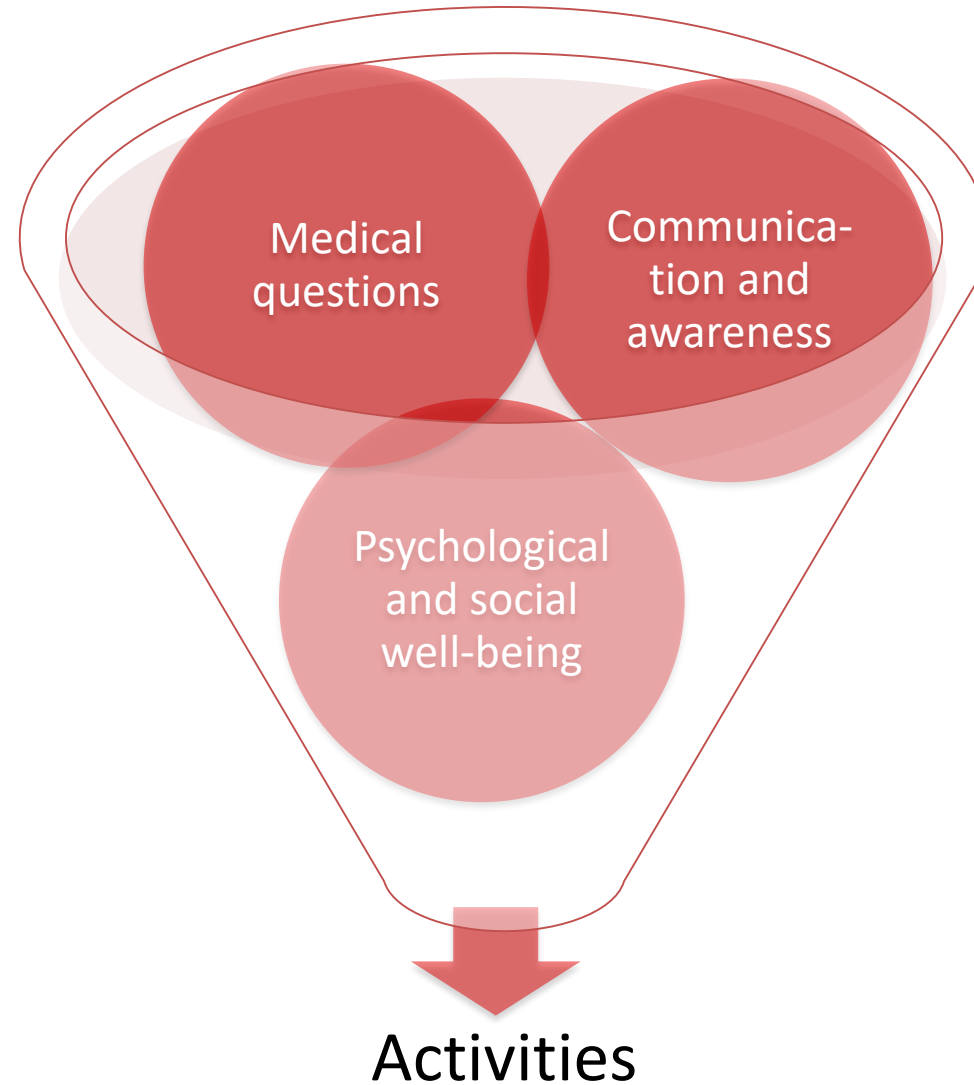
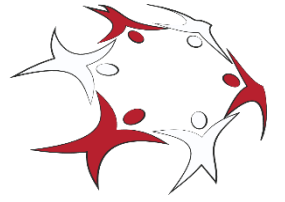
Women with at bleeding disorder



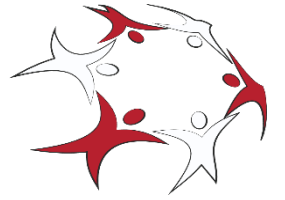
Symptoms:

- Frequent prolonged nose bleeds
- Prolonged bleeding from cuts
- Bleeding during and after surgery
- Easy bruising
- Heavy menstrual periods / menorrhagia

EHC Women working group



To recap



Recommendation 6

- **Genotype analysis** central to **understand** the disorder and **improve treatment**.
- **Genetic counselling** central for **family planning**.



Thank you for listening