特別プログラム

【学術集会企画】

■ Special Lecture

6 月 29 日（金） 11:10 ～ 12:00 第 1 会場（3F ロイトンホール AB）

Special Lecture 1 **座長：嶋 緑倫**（奈良県立医科大学小児科）

1-1

Family History for Hemophilia is negative at diagnosis in the majority of new children with severe hemophilia A ; Data from the PedNet registry.

PedNet Haemophilia Research Foundation **H. Marijke van den Berg**

1-2

Inhibitor incidence in **1083** PUPs with severe haemophilia A treated with class **Recombinant** or with class **Plasma-derived** products is similar; Recent data from the PedNet study group

H. Marijke van den Berg, on behalf of the PedNet study group.

Special Lecture 1-1

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Introduction

Haemophilia is an inherited rare disease occurring in 1: 5000 males. Data from cross-sectional surveys generally suggest that the family history for haemophilia is negative in 30% of the patients. In these patients, the diagnosis is only made after the onset of bleeding. Inhibitor risk is high during the first 50 exposure days (EDs) and for mitigating the risk especially the first 10-20 EDs are essential. Therefore, only patients diagnosed before the first bleeding might potentially be included in prospective clinical studies.
The aim of this study was to establish the number of PUPs with severe haemophilia A and a positive family history at the time of diagnosis, who as a consequence can be diagnosed before bleeding.

Methods and results

The PedNet study group (the European Paediatric Network for Haemophilia Management) is a collaboration of presently 33 haemophilia treatment centres (HTCs) in 17 countries, including Canada (Toronto and Montreal) and Israel. Data of well-defined clinical parameters are collected through a secured database system (www.pednet.eu). Data on the date of diagnosis and the family history at diagnosis is collected through the baseline forms. For this study the data of the January 2018 download were used.

Severe haemophilia A (factor VIII <0.01 IU/ml)	
Number of patients	1083
Family history at diagnosis **positive** for haemophilia	494 (46%)
Family history **positive**: age in years (IQR) at diagnosis	0.2 (0.0-0.1)
Family history **negative**: age in years (IQR) at diagnosis	0.8 (0.3-1.0)

Conclusion

The family history for haemophilia was positive in only 46% of the PUPs with severe haemophilia A; **54%** of the newly diagnosed severe haemophilia A patients have a negative family history. This has a large impact on strategies to prevent inhibitor development in all newly diagnosed PUPs with severe haemophilia A.

Special Lecture 1-2

Inhibitor incidence in **1083** PUPs with severe haemophilia A treated with class **Recombinant** or with class **Plasma-derived** products is similar; Recent data from the PedNet study group

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Introduction. The PedNet study group has published similar inhibitor incidence for rcFVIII versus pdFVIII in almost 600 PUPs with severe haemophilia A in the RODIN study based on data until June 2011. Now almost 500 more patients are included and 7-year more follow-up periods.

Objective. To investigate inhibitor incidence according to class FVIII type in a large unselected group of severe haemophilia A patients followed in 33 HTCs until January 2018.

Patients and methods. All patients with severe haemophilia A in the PedNet registry born after 1-1-2000 were included. Inhibitors were considered present if confirmed by at least two positive inhibitor assays. High-titre inhibitors were defined as an inhibitor titre >5 BU/ml. Patients were compared according to concentrate-type used at ED1: rcFVIII vs pdFVIII.

Results. A total of 1083 PUPS with severe haemophilia A were registered and 988 PUPs had received at least one exposure day. In 805 patients rcFVIII was used and 183 patients used pdFVIII. Follow up was complete at 50EDs for 95% on rcFVIII and 90% on pdFVIII. A total of 10 rcFVIII products and 15 pdFVIII products were used. Inhibitor incidence was similar across FVIII type used: for rcFVIII 29% (with 21% high titre) and for pdFVIII 28% (with 20% high titre). Patients that used pd products had more peak treatments at ED 1 than patients on rc products.

Conclusions

Similar risk for inhibitors between class rcFVIII and class pdFVIII was found in a large unselected cohort that was prospectively followed over 18 years. Analysis adjusted for individual genetic and treatment related risk factors (including individual products) is ongoing.