National study of inherited platelet function disorders in the Netherlands

Published: 29-12-2015
Last updated: 29-04-2024

Primary objective: to register and investigate Dutch patients suspect for an inherited platelet function disorder, to assess clinical presentation, bleeding score, treatment, burden of disease and quality of life. Secondary objectives: to...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Platelet disorders
Study type	Observational invasive

Summary

ID

NL-OMON54579

Source
ToetsingOnline

Brief title
Trombocytopathy in the Netherlands / TIN

Condition

- Platelet disorders
- Blood and lymphatic system disorders congenital

Synonym
trombocytopathy; platelet function disorders

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Utrecht
Source(s) of monetary or material Support: Ministerie van OC&W,deels door NWO Symphony project (NWA.1160.18.038),Sanofi-aventis,unrestricted grant van Sanquin Research

Intervention

Keyword: clinical features, diagnostic tests, inherited platelet function disorders

Outcome measures

Primary outcome

Frequency and severity of bleeding symptoms: bleeding score using the ISTH-BAT

Treatment of bleeding diathesis: type and frequency of treatment received in

the past (local treatment, antifibrinolytics, DDAVP, platelet transfusion)

Impact of PFD on quality of life, using age specific questionnaires.

Bruising pattern in children with PFD and VWD type 1 and 2

Secondary outcome

To investigate if diagnostic approaches can be improved and optimized using

tests additional to the standard available diagnostic tests available.

To search for a possible relationship between type of PFD and bleeding

phenotype.

To study phenotype-genotype relationships

To study occurrence and genotype-phenotype relationships within families

To gain more insight in pathophysiology of proven PFD*s

To validate a new disease-specific quality of life quiestionnaire

Study description

Background summary

Inherited platelet function disorders (PFD) are rare bleeding disorders caused by genetic defects, resulting in dysfunction of adhesion, activation or aggregation of platelets. This group of disorders is heterogeneous in severity, mechanisms and frequency. In the Netherlands little is known about prevalence,

clinical aspects, burden of disease and quality of life of patients with these disorders. An understanding of the impact of the disease, bleeding phenotype of the various kinds of PFD in the different age groups and improved diagnostics would help enormously in the early diagnosis, management and counseling of families with PFD and prevention of major bleedings.

Study objective

Primary objective: to register and investigate Dutch patients suspect for an inherited platelet function disorder, to assess clinical presentation, bleeding score, treatment, burden of disease and quality of life.
Secondary objectives: to investigate if diagnostic approaches can be improved and optimized using tests additional to the standard diagnostic tests. To search for a possible relationship between type of PFD and bleeding phenotype and to study genotype-phenotype relationships. To validate quality of life questionnaires in patients with PFDs.

Study design

Adults: Cross-sectional monocenter cohort study coordinated at the Van Creveldkliniek of the University Medical Center Utrecht.
Children: Corss-sectional mulitcenter study coordinated at the Van Creveldkliniek of the University Medical Center Utrecht in collaboration with the Erasmus Medical Center/Sophie Children's Hospital in Rotterdam and the Amsterdam University Medical Centre/Emma Children's Hospital Amsterdam.

Study burden and risks

This study will be the first to asses clinical presentation, quality of life and diagnostic approaches of patients with inherited PFDs in the Netherlands. The participating patients may benefit directly from participation as the use of new tests in this study may lead to diagnosing patients who were previously undiagnosed, resulting in better understanding and treatment options. The study consists of a questionnaire and the drawing of extra blood to perform additional tests. Risks imposed by participation are considered negligible.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)
Babies and toddlers (28 days-23 months)

Inclusion criteria

(Suspected) PFD defined according to the following criteria:
• Congenital or familial thrombocytopenia
• Chronic thrombocytopenia (>1 year) without proven or suspected acquired cause
• PFD with proven molecular diagnosis.
• Abnormal LTA for at least one of the agonists and/or abnormal ATP/ADP ratio (storage pool test)
• History of bleeding diathesis very suspect of a primary hemostasis function defect with or without prolonged Platelet Function Analyser closure time.

Inclusion criteria of control group of VWD patients for bruising pattern
Patients younger than 18 years old with Von Willebrand*s disease type 1 or type 2 that visit the VCK regularly.

Exclusion criteria

- Inability to give informed consent or inability of the parents to give

informed consent in patients < 16 years of age.
- Bleeding diathesis due to an acquired PFD
- Bleeding diathesis due to moderate or severe von Willebrand disease (VWF risto < 30%)
- Bleeding diathesis due to hemophilia or other disorders of secondary hemostasis or fibrinolysis
- Current use of antiplatelet therapy

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Diagnostic

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	11-02-2016
Enrollment:	600
Type:	Actual

Medical products/devices used

Registration:	No

Ethics review

Approved WMO Date:	29-12-2015
Application type:	First submission
Review commission:	METC NedMec
Approved WMO Date:	16-11-2016

Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	02-03-2018
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	09-05-2018
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	10-09-2021
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	16-11-2022
Application type:	Amendment
Review commission:	METC NedMec
Approved WMO Date:	07-06-2023
Application type:	Amendment
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)
Approved WMO Date:	17-04-2024
Application type:	Amendment
Review commission:	METC Universitair Medisch Centrum Utrecht (Utrecht)

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL53207.041.15