# Platelet function in 22q11.2 deletion syndrome

No registrations found.

| | |
|---|---|
| **Ethical review** | Not applicable |
| **Status** | Pending |
| **Health condition type** | - |
| **Study type** | Interventional |

## Summary

### ID

NL-OMON24347

**Source**
Nationaal Trial Register

**Brief title**
TBA

**Health condition**

22q11.2 deletion syndrome, schizophrenia, thrombocytopenia

### Sponsors and support

**Primary sponsor:** None
**Source(s) of monetary or material Support:** None

### Intervention

### Outcome measures

**Primary outcome**

- Bleeding risk score (ISTH-BAT questionnaire).
- Complete blood count.
- Platelet aggregation and (functional) flowcytometry.
- Flow chamber results with respect to platelet binding to coated surfaces (in bright field view), P-selectin expression, fibrinogen binding and phosphatidyl serine (PS) exposure.

- Global scale quantitative and qualitative RNA differences (transcriptomics).
- Global scale quantitative metabolite differences (metabolomics).

### Secondary outcome

Correlation between age and platelet function and platelet count.

## Study description

### Background summary

Background: 22q11.2 Deletion syndrome (22q11.2DS) is caused by recurrent heterozygous microdeletions on chromosome 22q11.2, encompassing up to 90 genes. This syndrome is characterized by a multi-organ disorder with a variable phenotype, including intellectual disability, cognitive deterioration, schizophrenia, early-onset Parkinson's disease, recurrent epistaxis, and macrothrombocytopenia; ~30% of the adults with 22q11.2DS have thrombocytopenia (<150,000 platelets per mL). Schizophrenia occurs in ~25% of individuals with 22q11.2DS, and ~1-2% of individuals with schizophrenia in the general population have the 22q11.2DS. Approximately 40% of the individuals with 22q11.2DS has intellectual disability.
Importantly, platelets have a critical role in hemostasis. Also, they show similarities to neurons concerning several morphologic and biochemical characteristics, are easier to investigate, and may therefore serve as a window to the brain.
Only a limited number of studies has investigated bleeding risk and platelet function in 22q11.2DS, and those who did only included children. Some of these studies indicated impaired platelet function and increased bleeding risk, and one reported a negative correlation between platelet count and age, which may suggest that platelet-associated problems increase with increasing age.

Aim: The combination of bleeding risk score and platelet function analysis with platelet transcriptomics and metabolomics may: 1) provide insight into bleeding risk, which is of direct relevance for patient care, and 2) provide insight in mechanisms underlying neurodevelopmental and neuropsychiatric disorders, like schizophrenia, that are frequently seen in 22q11.2DS.

Methods: we will include 40 adults with 22q11.2DS (20 with schizophrenia, 20 without schizophrenia) and 20 healthy controls. All participants will be assessed once (1 our in total), this includes blood drawl and completing a bleeding risk questionnaire.

### Study objective

1) there will be a significant difference in platelet function and bleeding risk between adults with 22q11.2DS and healthy controls
2) there will be differences in RNA and metabolites between 22q11.2DS with schizophrenia

and without schizophrenia as well as between 22q11.2DS in general and healthy controls

**Study design**

Cross-sectional, only one assessment

**Intervention**

Blood drawl and ISTH-BAT questionnaire

# Contacts

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# Eligibility criteria

## Inclusion criteria

In order to be eligible to participate in this study, a subject must meet all of the following criteria:
- 16 years or older.
- signed informed consent.

Adults with 22q11.2DS
- molecularly confirmed 22q11.2 deletion syndrome.
- Mentally competent (ability to give informed consent) and aged 16 years and older or, in case the individual is mentally incompetent aged 16 years and older, consent will be given by the legally authorized representative of the subject.

### Exclusion criteria

- The presence of any malignancy.
- Use of antiplatelet or anticoagulant drugs within the last two weeks prior to the study.
- Use of anti-inflammatory drugs within the last two weeks prior to the study.
- Medical history of auto-immune thrombocytopenia

Specific for healthy controls:
- A medical history of thrombocytopenia (<150.000 platelets per mL).
- Increased bleeding risk, defined as a diagnosed bleeding disorder.
- Metabolic disorder.

## Study design

### Design

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|---|---|
| Study type: | Interventional |
| Intervention model: | Other |
| Allocation: | Non controlled trial |
| Masking: | Open (masking not used) |
| Control: | Active |

### Recruitment

NL

| | |
|---|---|
| Recruitment status: | Pending |
| Start date (anticipated): | 01-05-2021 |
| Enrollment: | 60 |
| Type: | Anticipated |

### IPD sharing statement

**Plan to share IPD:** Undecided

## Ethics review

Not applicable

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|---|---|
| Application type: | Not applicable |

# Study registrations

## Followed up by the following (possibly more current) registration

ID: 55192
Bron: ToetsingOnline
Titel:

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

No registrations found.

## In other registers

| Register | ID |
|---|---|
| NTR-new | NL9363 |
| CCMO | NL75078.068.21 |
| OMON | NL-OMON55192 |

# Study results