

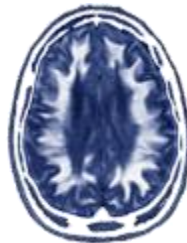
The MLD initiative: an academia-led registry for metachromatic leukodystrophy

Daphne Schoenmakers, MD PhD student

27-9-2021



Amsterdam
Leukodystrophy
Center



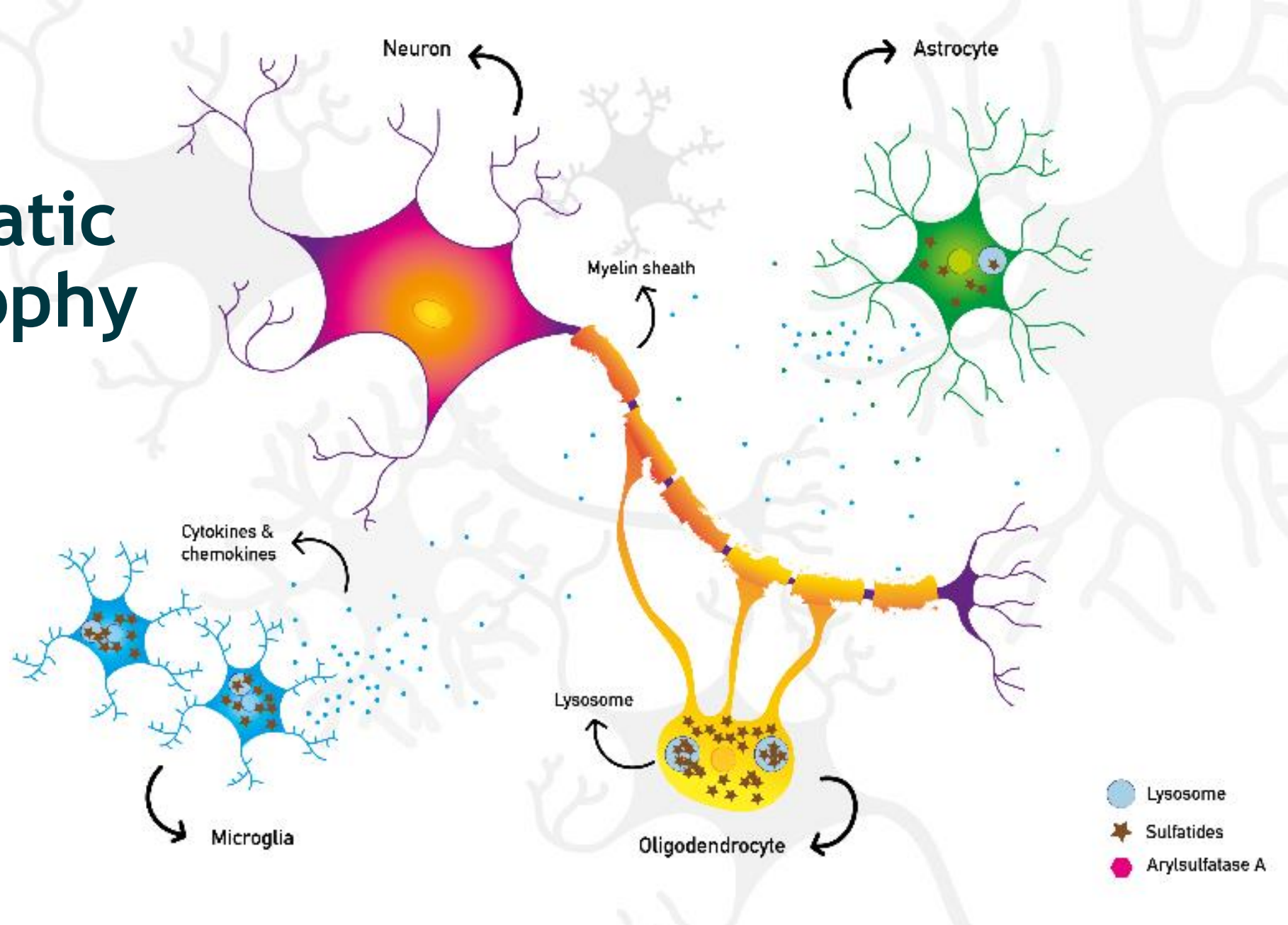
Medicijn
voor de
maatschappij



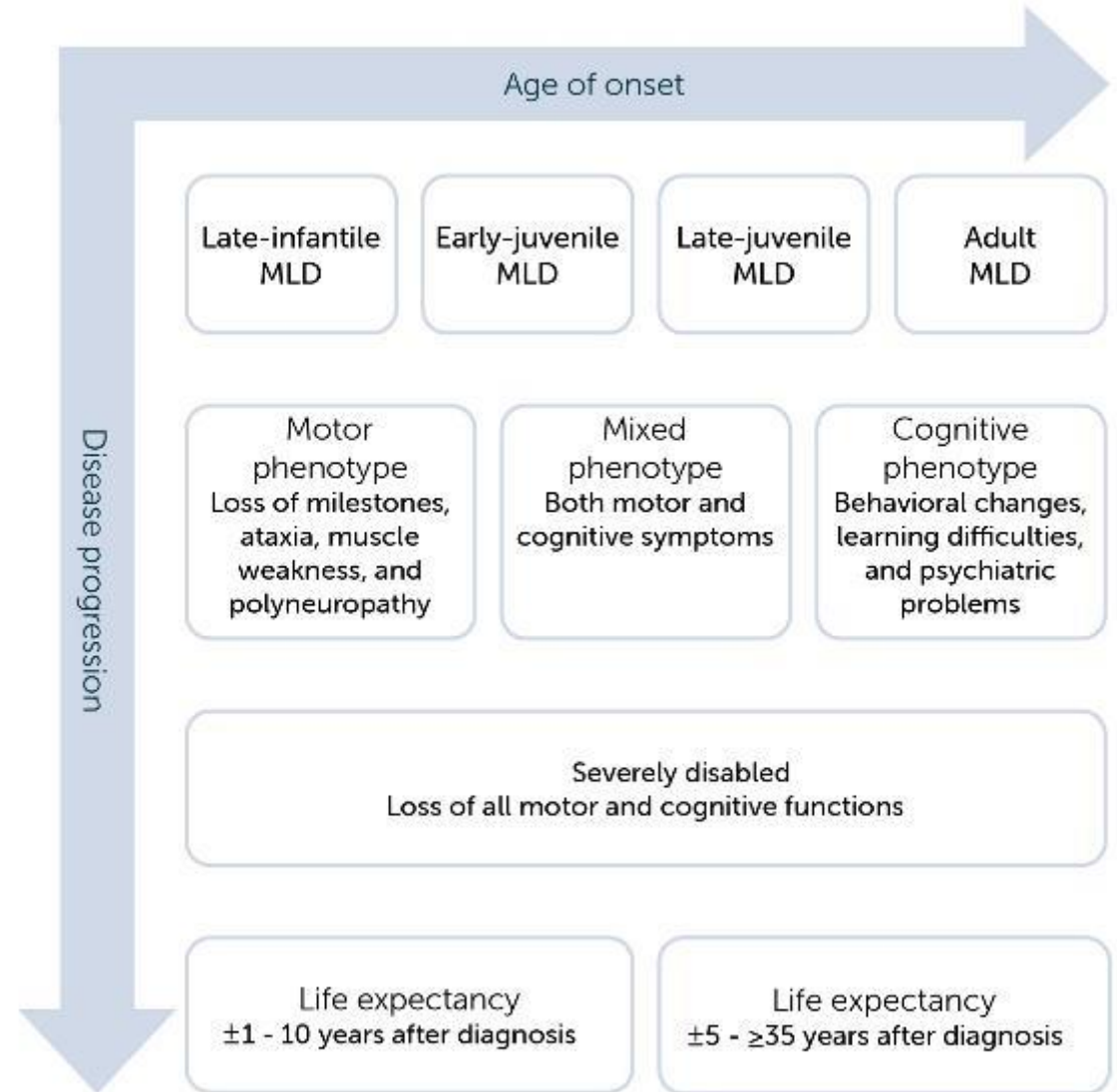
Zorginstituut Nederland
RORDGM

VRIENDENLOTERIJ

What is Metachromatic leukodystrophy (MLD)?



What are the symptoms of MLD?





What are the therapeutic options?

Supportive care

Hematopoietic stem cell transplantation

Autologous HSCT-based gene therapy (Libmeldy)

Intrathecal enzyme replacement therapy (phase 2 trial)



What are the therapeutic options?

Supportive care

Hematopoietic stem cell transplantation

Autologous HSCT-based gene therapy (Libmeldy)

Intrathecal enzyme replacement therapy (phase 2 trial)



Additional monitoring

- Additional monitoring
- Post-authorisation evaluation
- Industry-led registry for specific drug



EUROPEAN MEDICINES AGENCY
SCIENCE MEDICINES HEALTH

Libmeldy [Share](#) [RSS](#)

Autologous CD34+ cells encoding ARSA gene

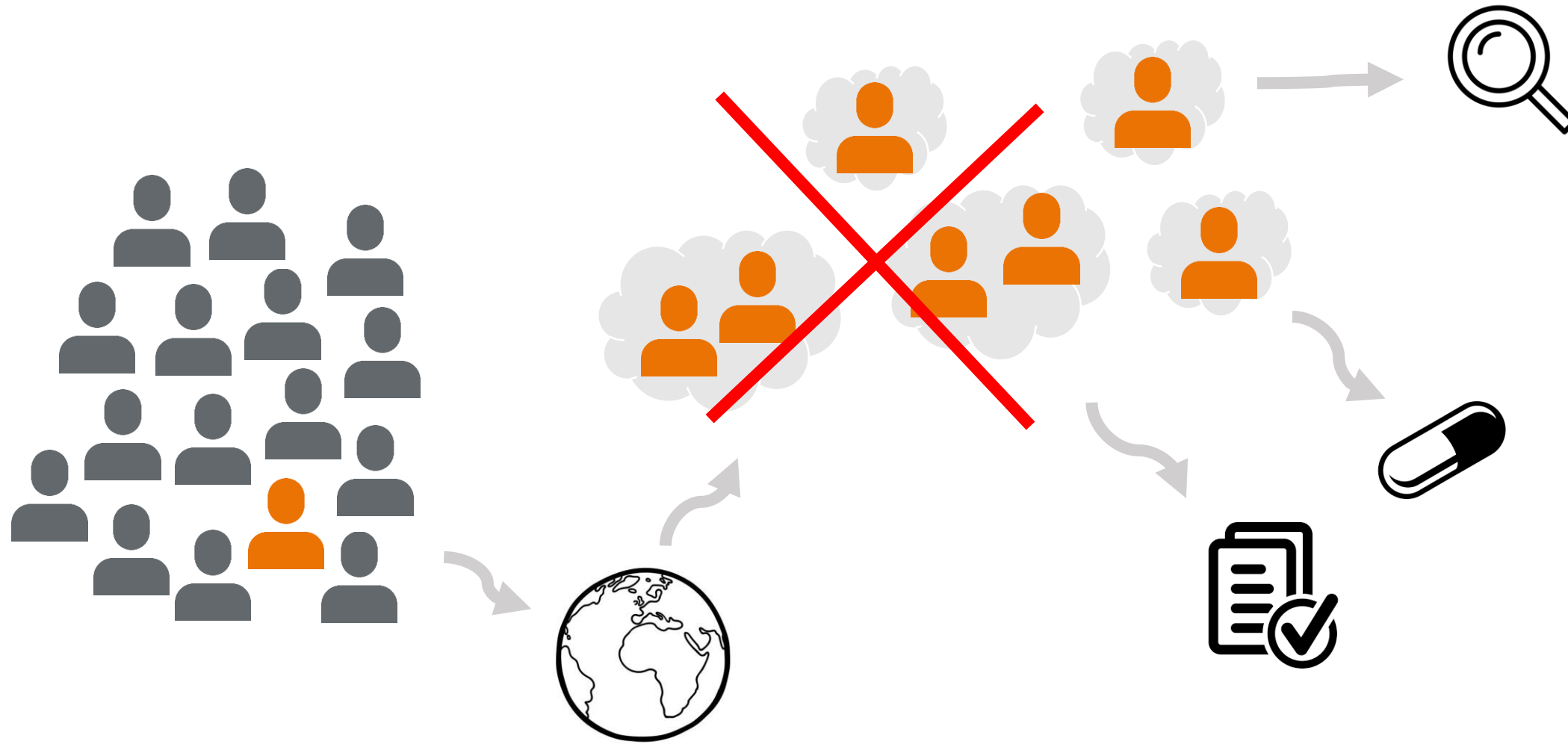
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AUTHORISED

This medicine is authorised for use in the European Union.

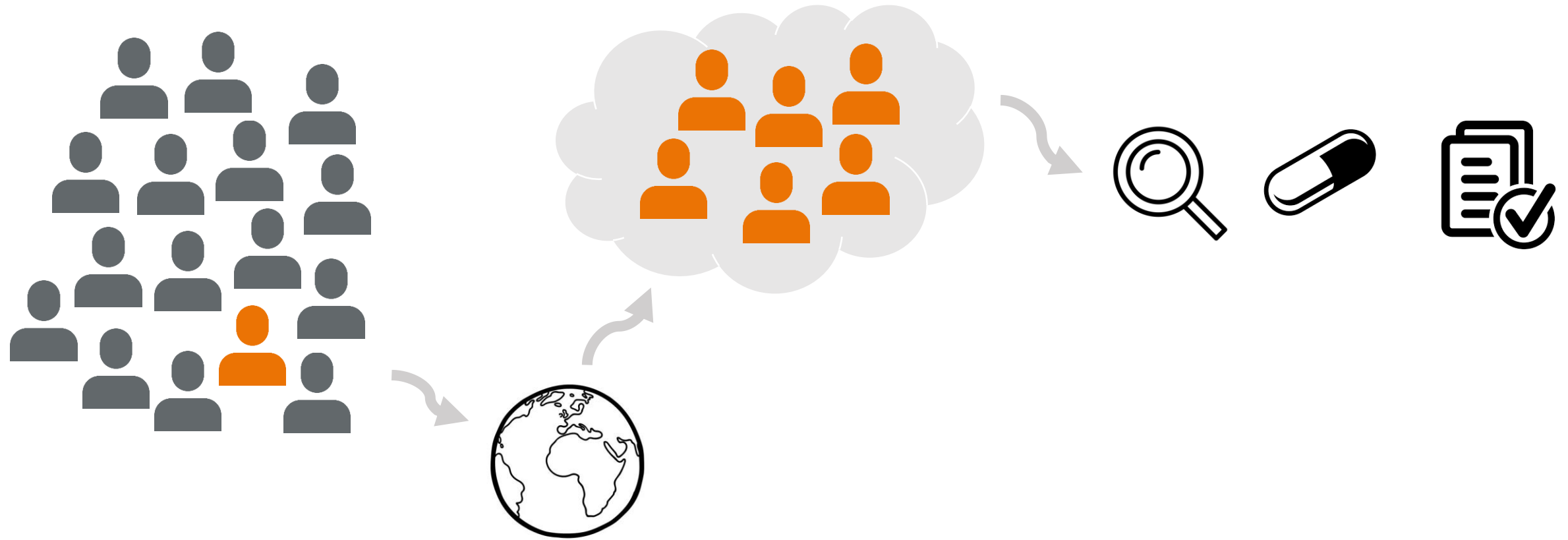


Background

MLDi

Triangle

Readiness



Background

MLDi

Triangle

Readiness



*To improve disease management of metachromatic leukodystrophy
through an international disease registry and multi-stakeholder collaboration.*

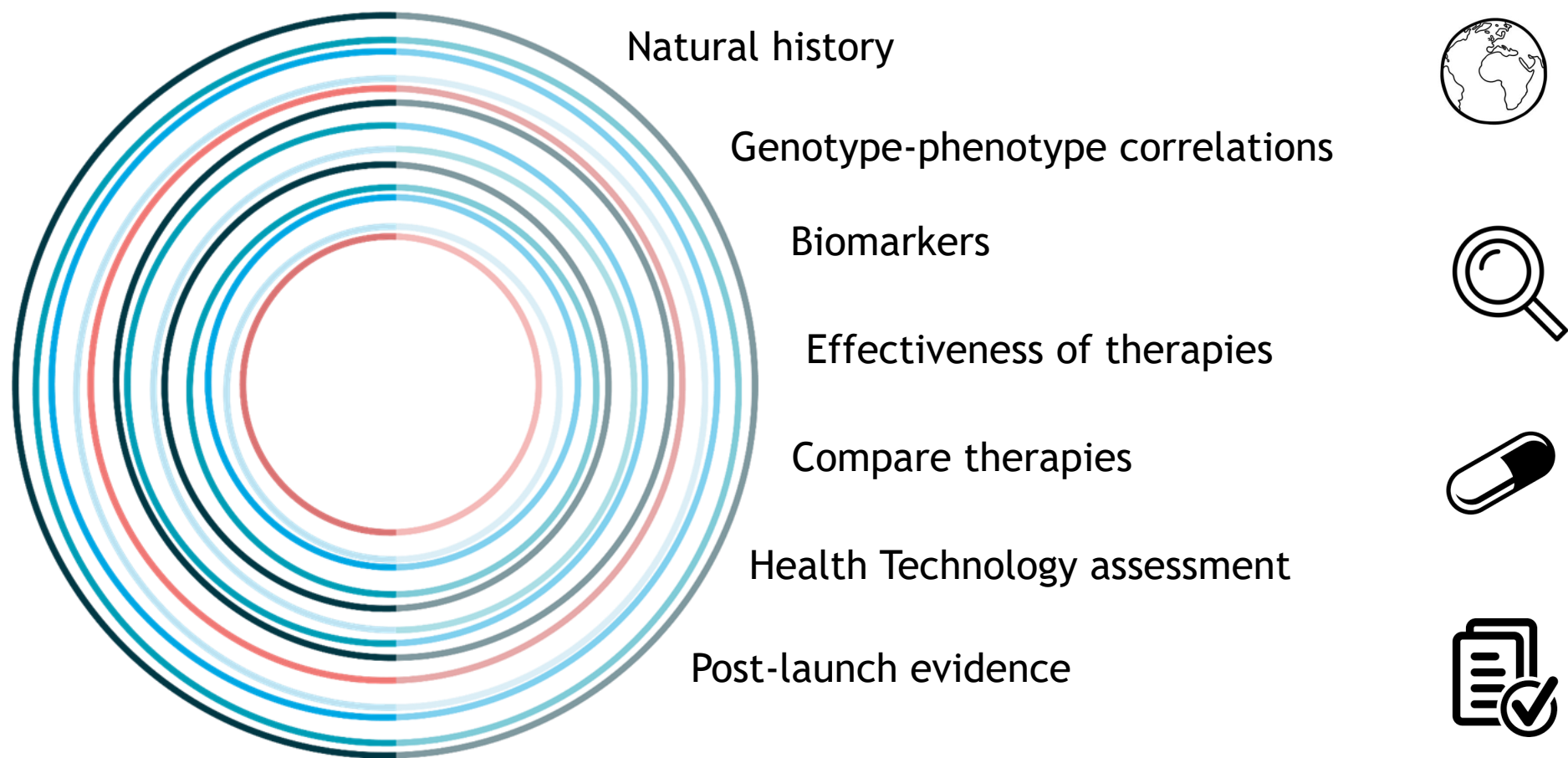


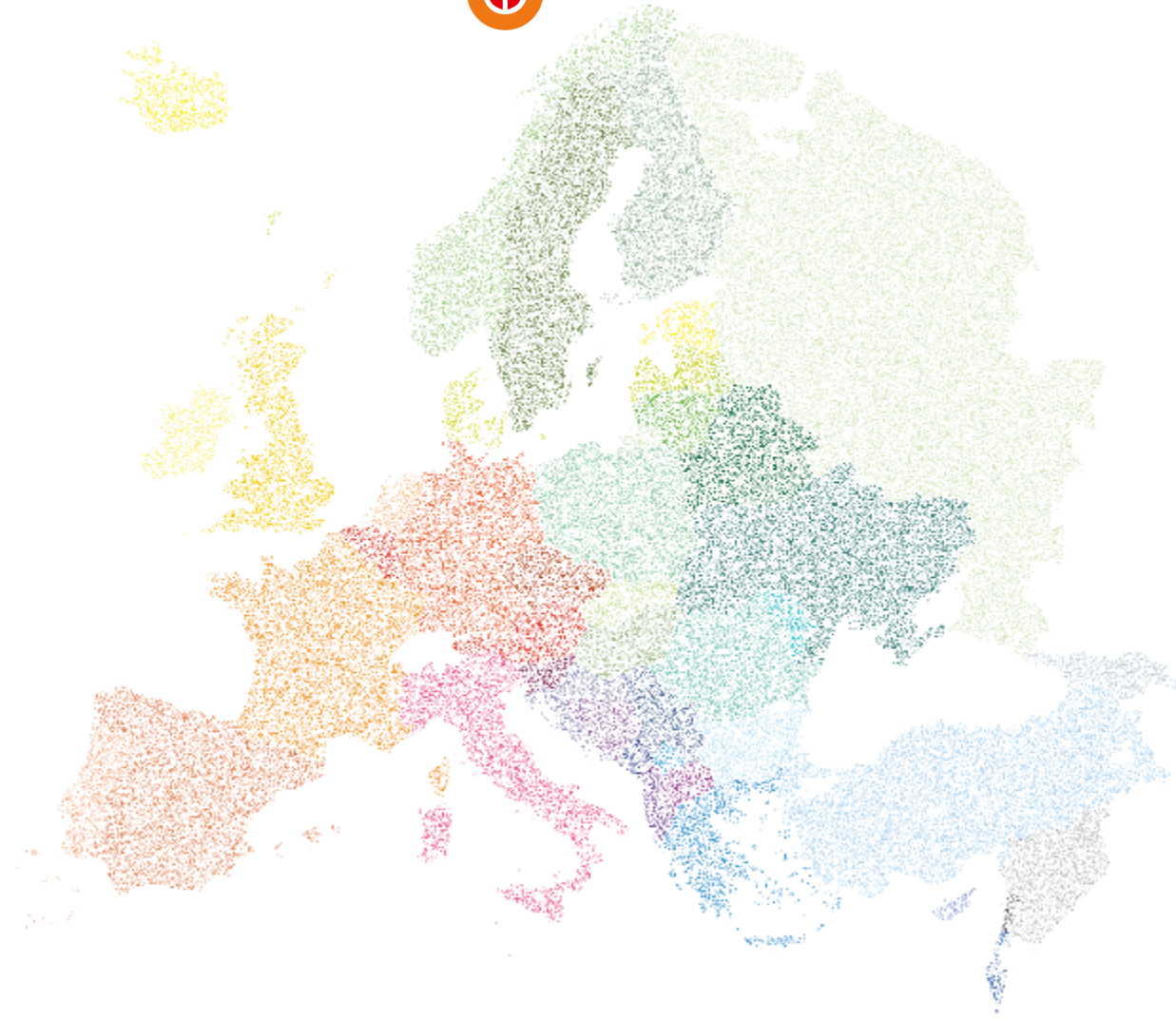
the MLD initiative





One academia-led international multi-purpose disease registry for MLD



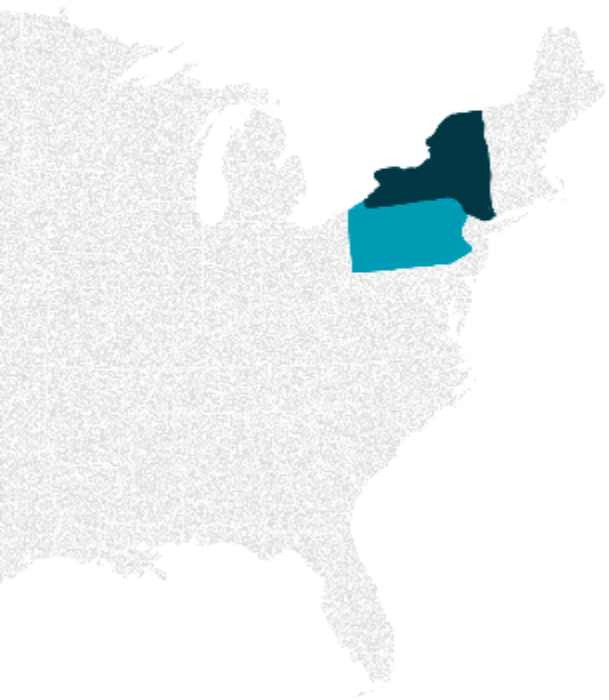


Background

MLDi

Triangle

Readiness



Dr. Laura Adang, Philadelphia
Dr. Annette Bley, Hamburg
Prof. dr. Jaap-Jan Boelens, New York
Dr. Valeria Calbi, Milan
Prof. dr. Antonio Federico, Siena
Dr. Francesca Fumagalli, Milan
Dr. Sabine Grønberg, Copenhagen
Dr. Samuel Gröschel, Tübingen
Dr. Joanne Hughes, Dublin
Dr. Simon Jones, Manchester
Dr. Caroline Lindemans, Utrecht
Dr. Fanny Mochel, Paris
Dr. Dipak Ram, Manchester
Prof. dr. Ludger Schöls, Tübingen
Dr. Caroline Sevin, Paris
Dr. Peter van Hasselt, Utrecht
Prof. dr. Nicole Wolf, Amsterdam
Prof. dr. Robert Wynn, Manchester
Dr. Ayelet Zerem, Tel Aviv

19 experts
15 expert centers
9 countries

Background

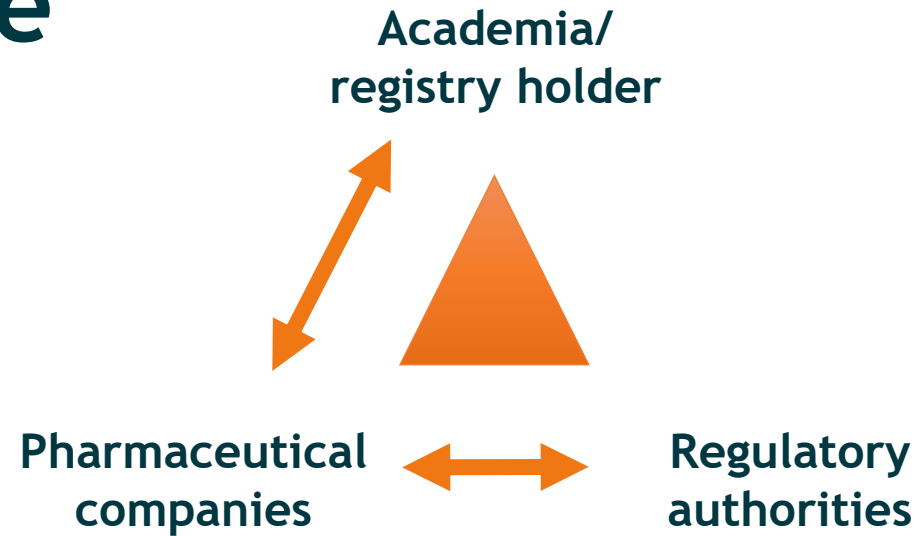
MLDi

Triangle

Readiness



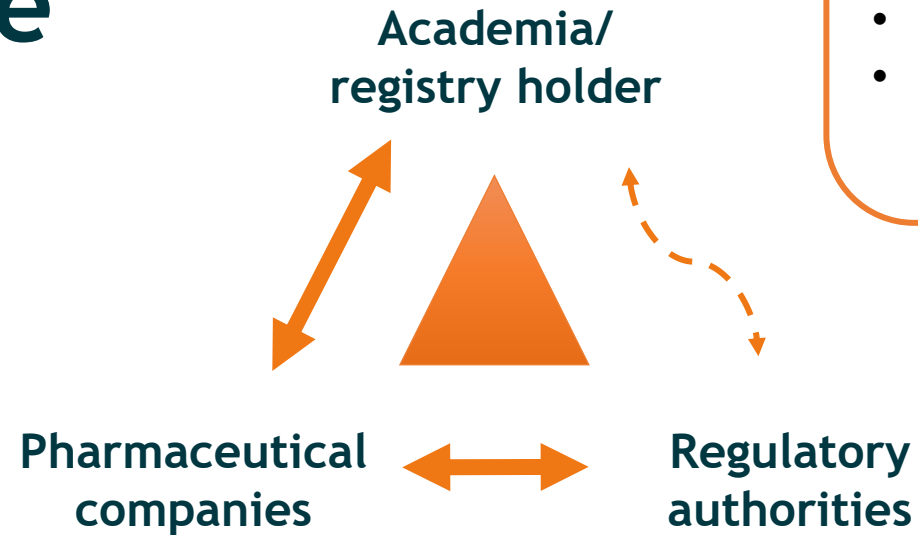
Broken triangle



Source: Nicole Ruperto, PRINTO; EMA patient registries initiative workshop



Broken triangle

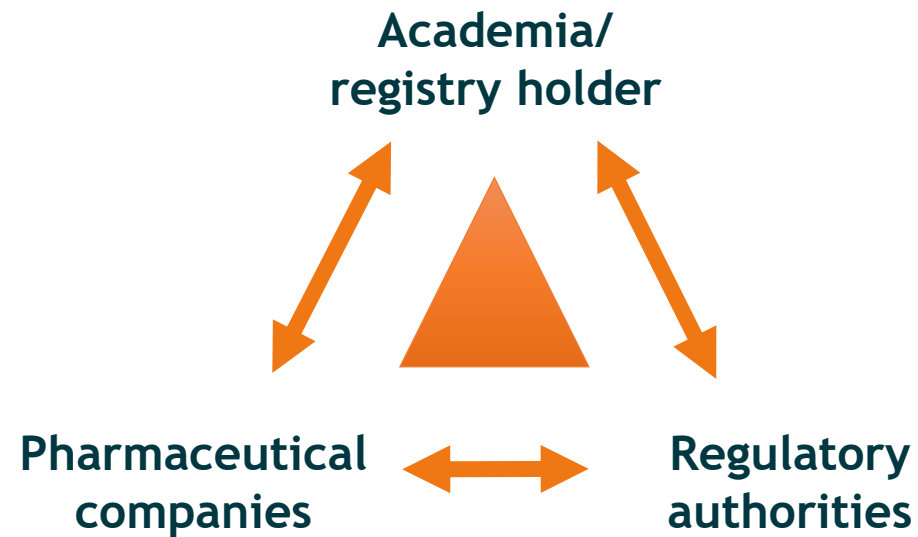


- EU-RD Platform
- EMA guideline on registry-based studies (DRAFT)
- REQueST tool (EUnetHTA)
- EUnetHTA PLEG activities
- EMA qualification for ECFSPR & EBMT

Source: Nicole Ruperto, PRINTO; EMA patient registries initiative workshop



Fixed triangle



Zorginstituut Nederland

"Managing patient
registries for
expensive drugs" /
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✓ **AUTHORISED**
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 Benelux
Initiative on Pharmaceutical Policy





How ready are we?

- Collaborative network
- High quality data:
 - Modified Delphi consensus procedure
- Input from stakeholders
 - ZIN, EMA, CBG, EU-RD-platform/EC, ERNs, patients & families, patient advocacy groups
- IT infrastructure
- Governance
- Fee waiver for EMA qualification opinion procedure



Future

- Expand data collection
- RORDGM
- EMA Qualification opinion
- Learning-by-doing & learning from others
- Share experiences and best-practices
- Involvement and alignment of stakeholders



*To improve disease management of metachromatic leukodystrophy
through an international disease registry and multi-stakeholder collaboration.*



the MLD initiative





Prof. Dr. Nicole I.
Wolf, child neurologist



Prof. Dr. Carla E.M. Hollak, internist
inherited metabolic diseases



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Drs. Shanice
Beerepoot
MD, PhD student



Drs. Daphne
Schoenmakers
MD, PhD student



Drs. Sibren van de Berg
Coördinator Medicijn voor de
Maatschappij, PhD student



Drs. Mareen Datema
Datamanager

