

Intro to the brain and behavior: the “why” behind learning & behaviors

**Parent
Project
Muscular
Dystrophy**

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Why neuroimaging in DMD & BMD?

To study the consequences of dystrophinopathy in the brain



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Neuroimaging in DMD & BMD

What is magnetic resonance imaging (MRI)?

- MRI scanner: strong magnetic field

Large scanner
3 Tesla =
= 52,000 x earth
magnetic field



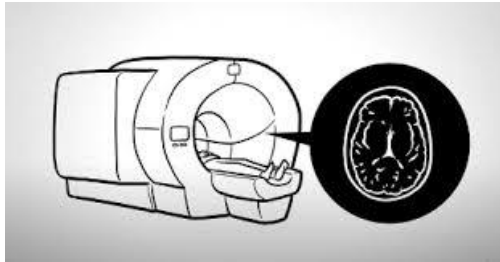
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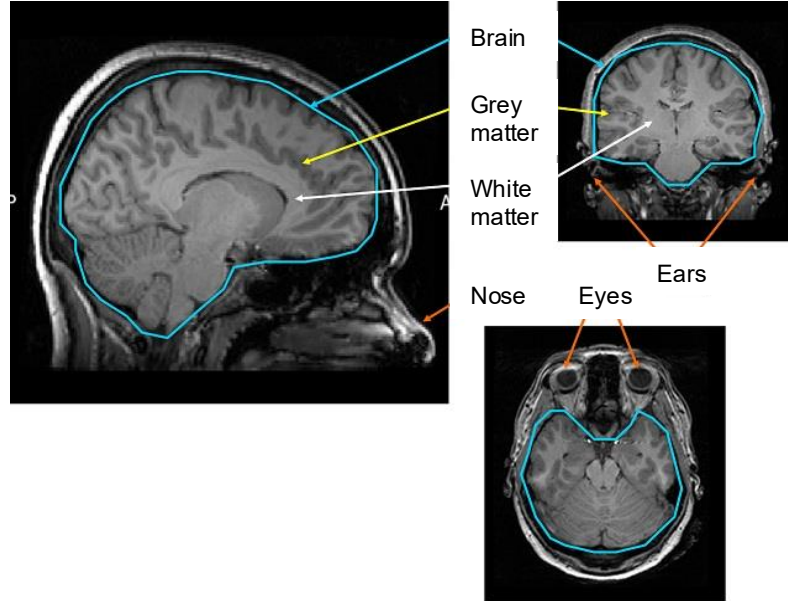
Neuroimaging in DMD & BMD

What is magnetic resonance imaging (MRI)?

- MRI scanner: strong magnetic field
- Look inside the brain
- Different types of scans



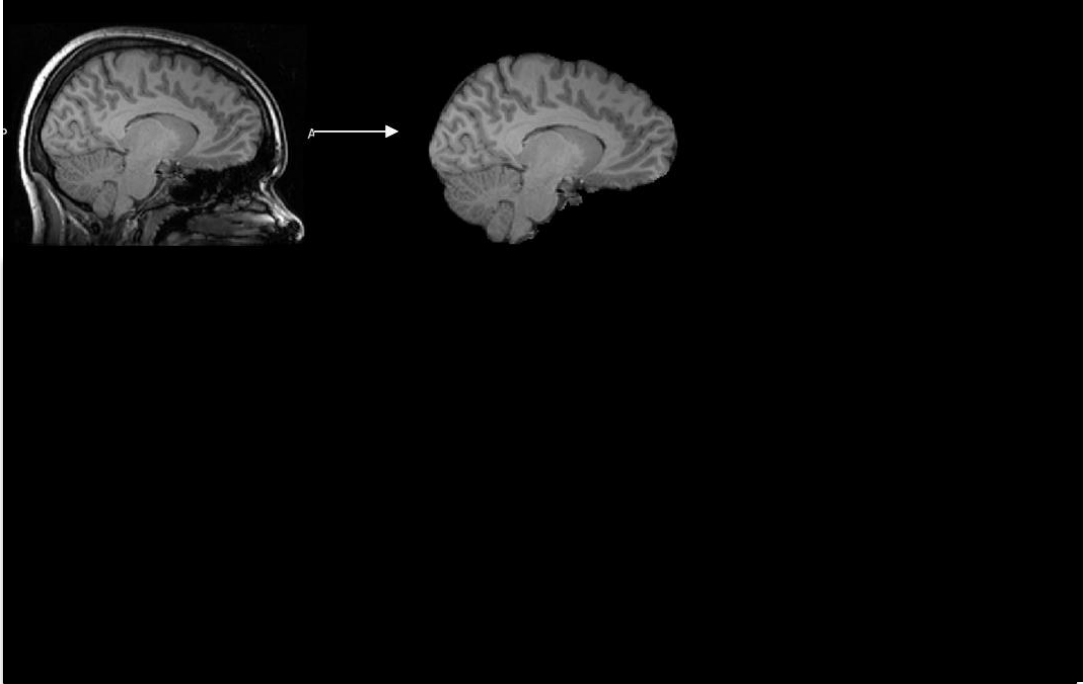
Neuroimaging in DMD & BMD



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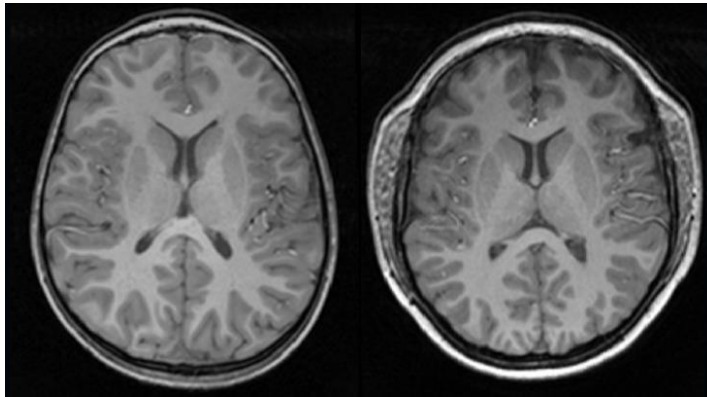
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DMD Anatomy (volumetrics)



Anatomy (volumetrics)

- Unknown in BMD
- In DMD:
 - No gross anatomical changes
 - Smaller total brain, grey matter, white matter, and cerebrospinal fluid volumes
 - Spread throughout the brain > no clear localized effect
 - Temporal muscle hypertrophy and a more circular head shape



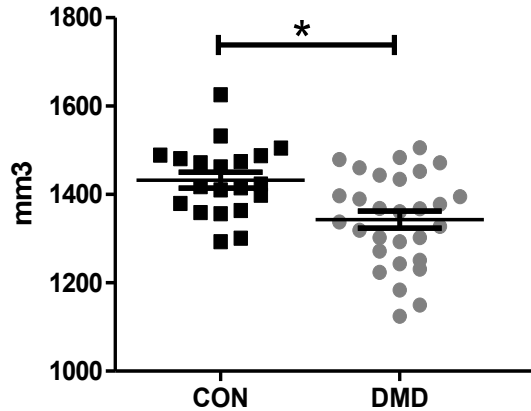
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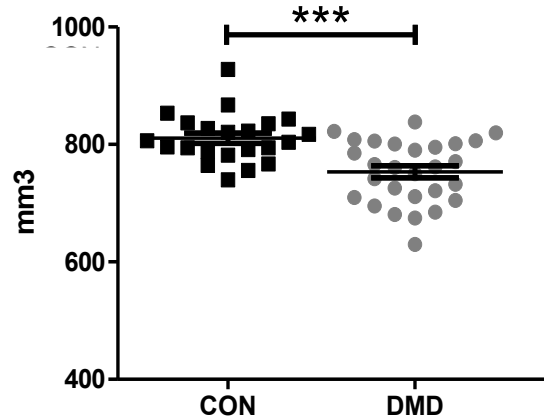
*Straathof, J. Child. Neurol. 2014
Doorenweerd, Ann. Neurol. 2014
Geuens, Ann Clin Transl Neurol. 2023*

DMD Anatomy (volumetrics)

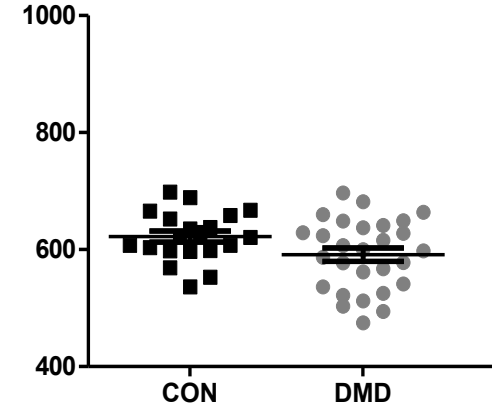
Total brain volume



Grey matter

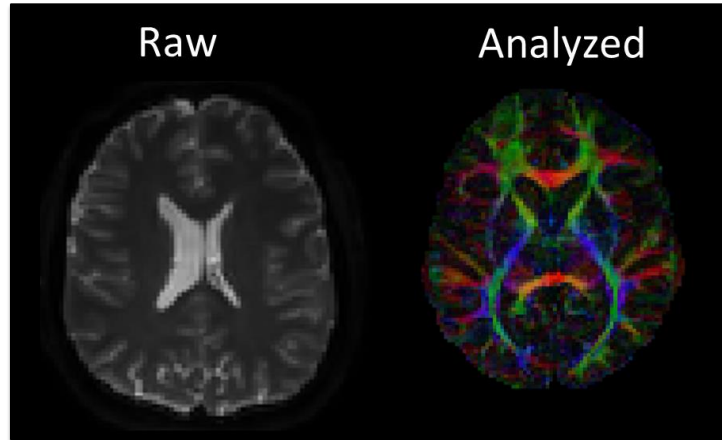
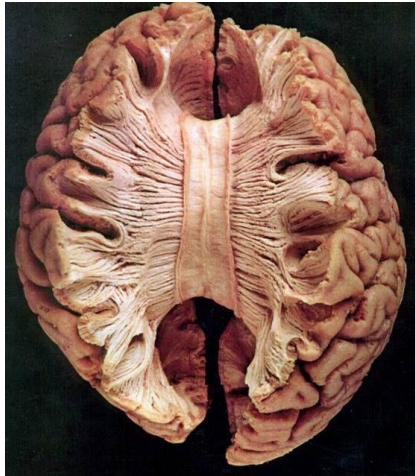


White matter



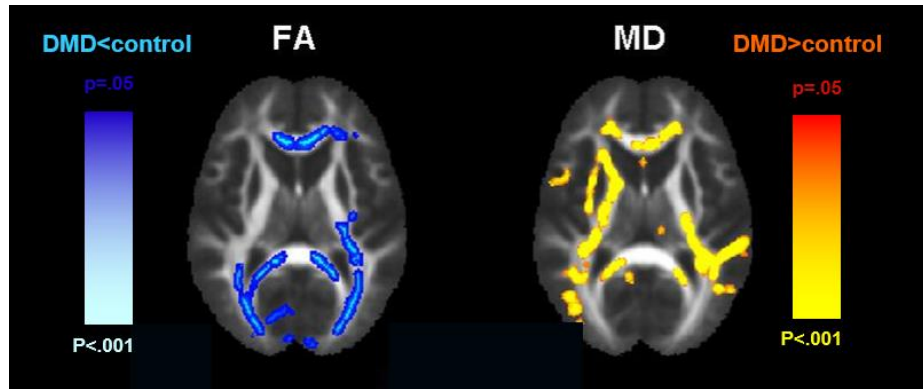
Microstructure (diffusion)

- 'Highways' of the brain



Microstructure (diffusion)

- Unknown in BMD
- In DMD:
 - Different white matter microstructure
 - Spread throughout the white matter > No clear localized effect



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Doorenweerd, *Ann. Neurol.* 2014
Fu, *Ann Transl Med.* 2016
Preethish-Kumar, *AJNR Am J Neuroradiol.* 2020
Biagi, *PLoS One.* 2021
Govaarts, *NMR Biomed.* 2024

DMD Imaging & Cognition

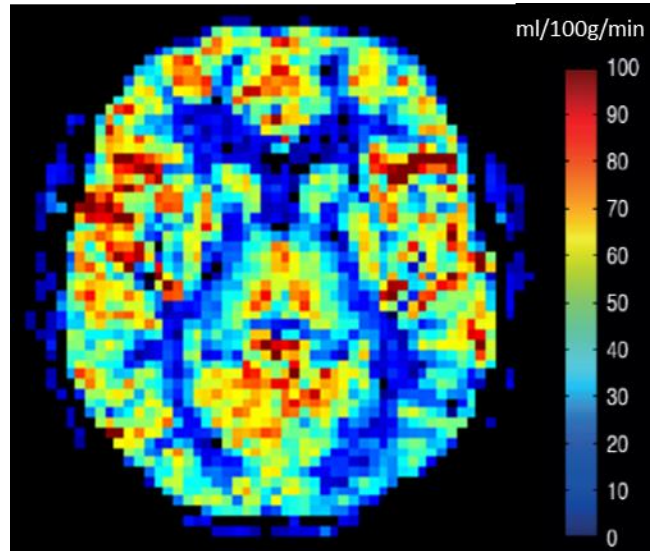
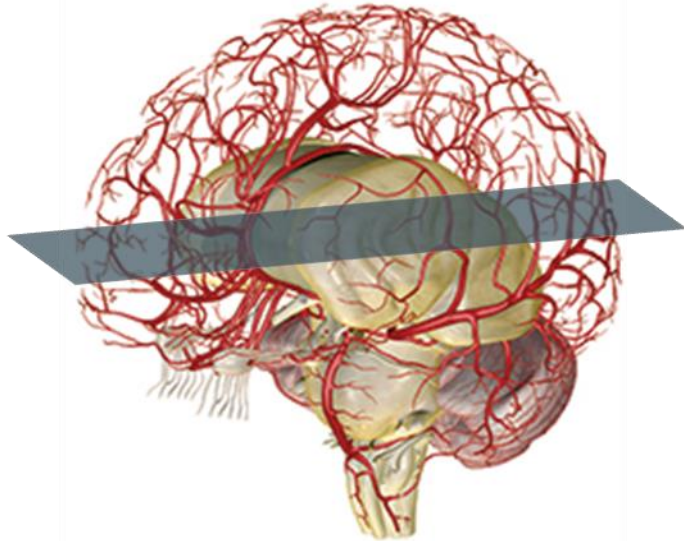
- Microstructure (diffusion) shows a correlation with:
 - Full Intellectual Quotient (WISC-IV)
 - Denomination (Naming) (total score, Nepsy II)
 - Inhibition (total score, Nepsy II)



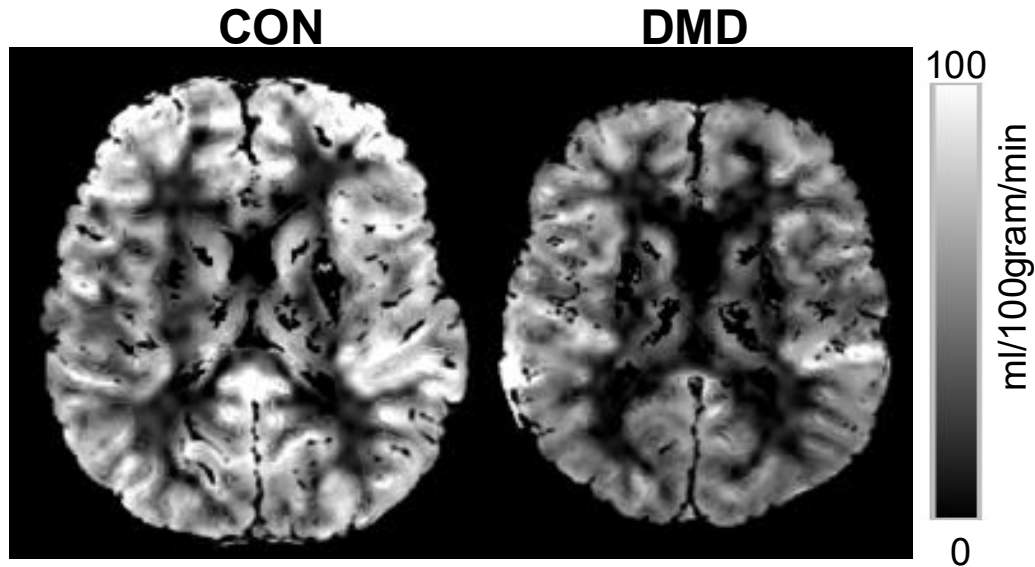
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Cerebral blood flow (perfusion)

- How much blood is flowing through the brain



Cerebral blood flow (perfusion)



- Unknown in BMD
- In DMD:
 - Lower cerebral perfusion (by ~17%)
 - Spread throughout the grey matter > no clear localized effect
 - No correlation with grey matter volume

Summary

- In **BMD**, there are **no clear differences** in brain volumetrics, microstructure, and perfusion
- In **DMD**, there are **clear imaging correlates** with dystrophinopathies
 - Larger groups are needed to link cognitive and behavioral functioning to brain imaging



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Acknowledgement

Many thanks to the participants and their families!

Team of the Leiden University Medical Center



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Thank you!



Questions?