Dopamine in 22q11

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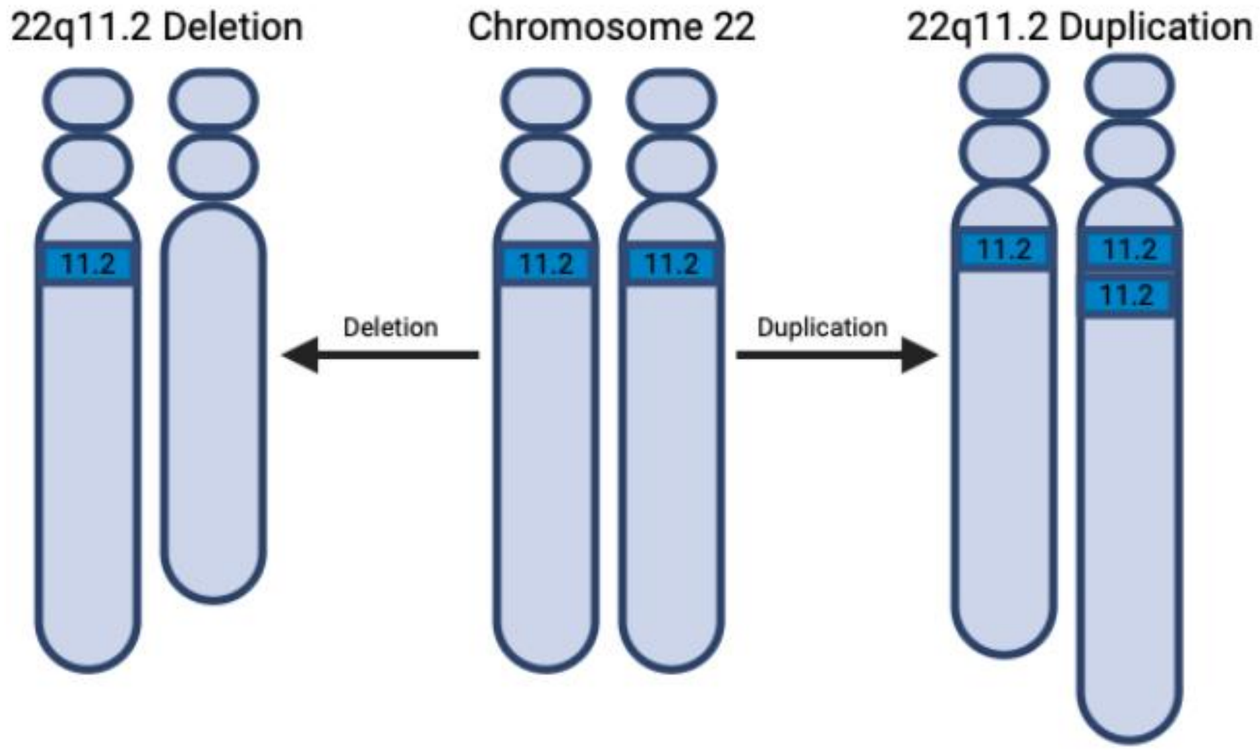
Research experience

3 years with 22q11 CNV

First year
- Meeting patients and their familiesat their home
- 16-80 years old

Last two years
- Brain research

22q11.2 copy number variants



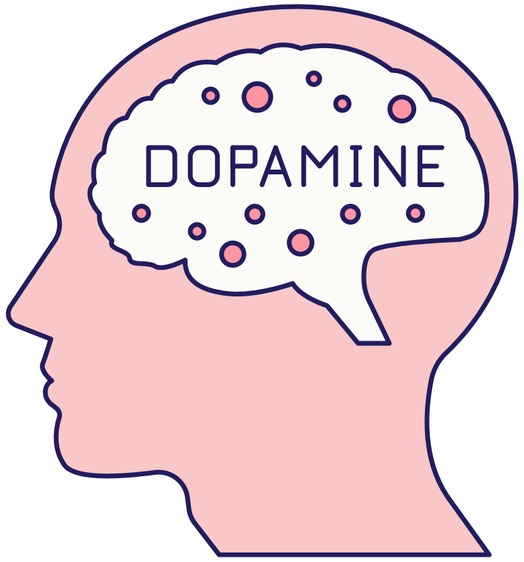
This image is created in BioRender.

22q11.2 CNV

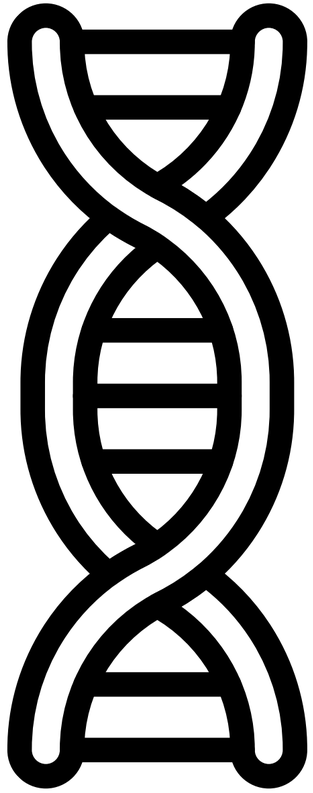
22q11 deletion and 22q11 duplication both have a higher risk of developing:

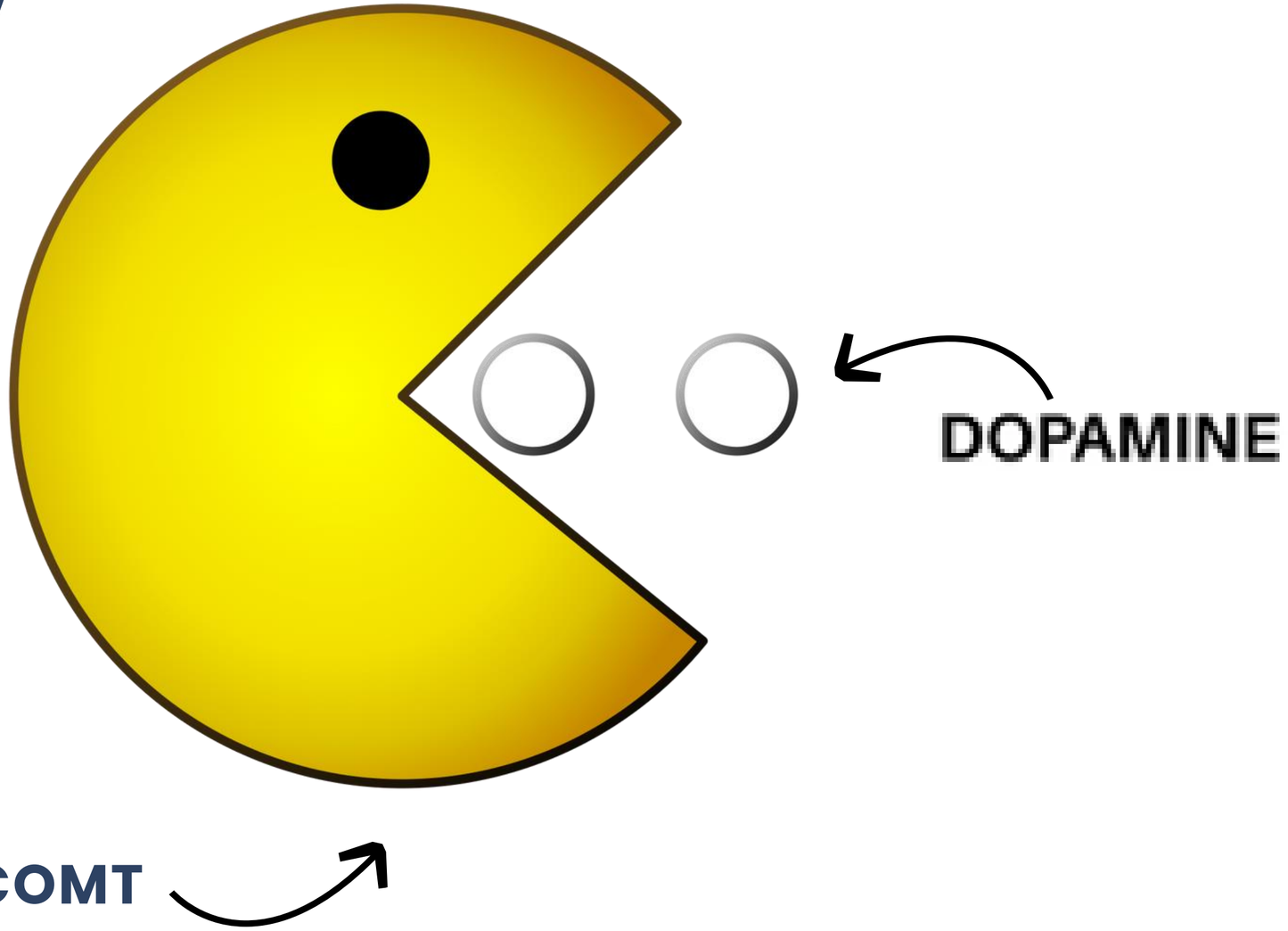
- ADHD
- Autism
- Learning difficulties
- Mood problems

Dopamine

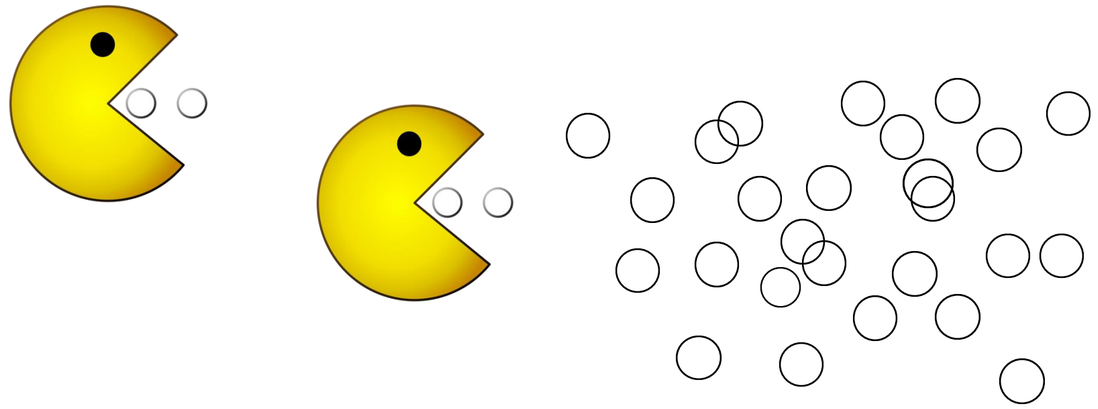


22q11 CNV

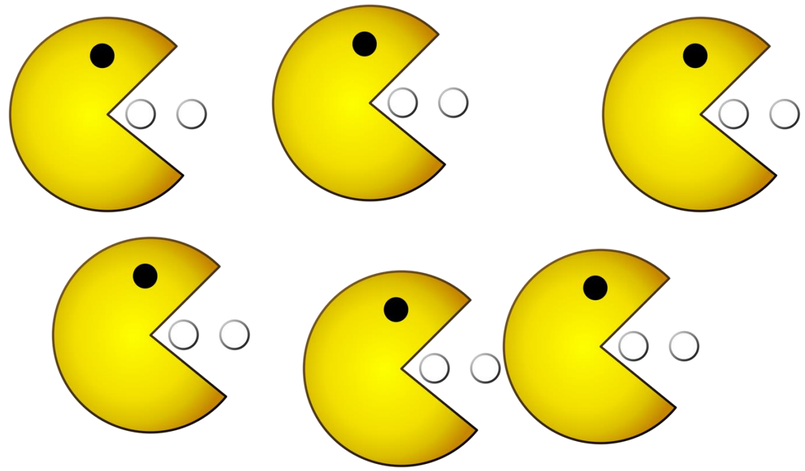




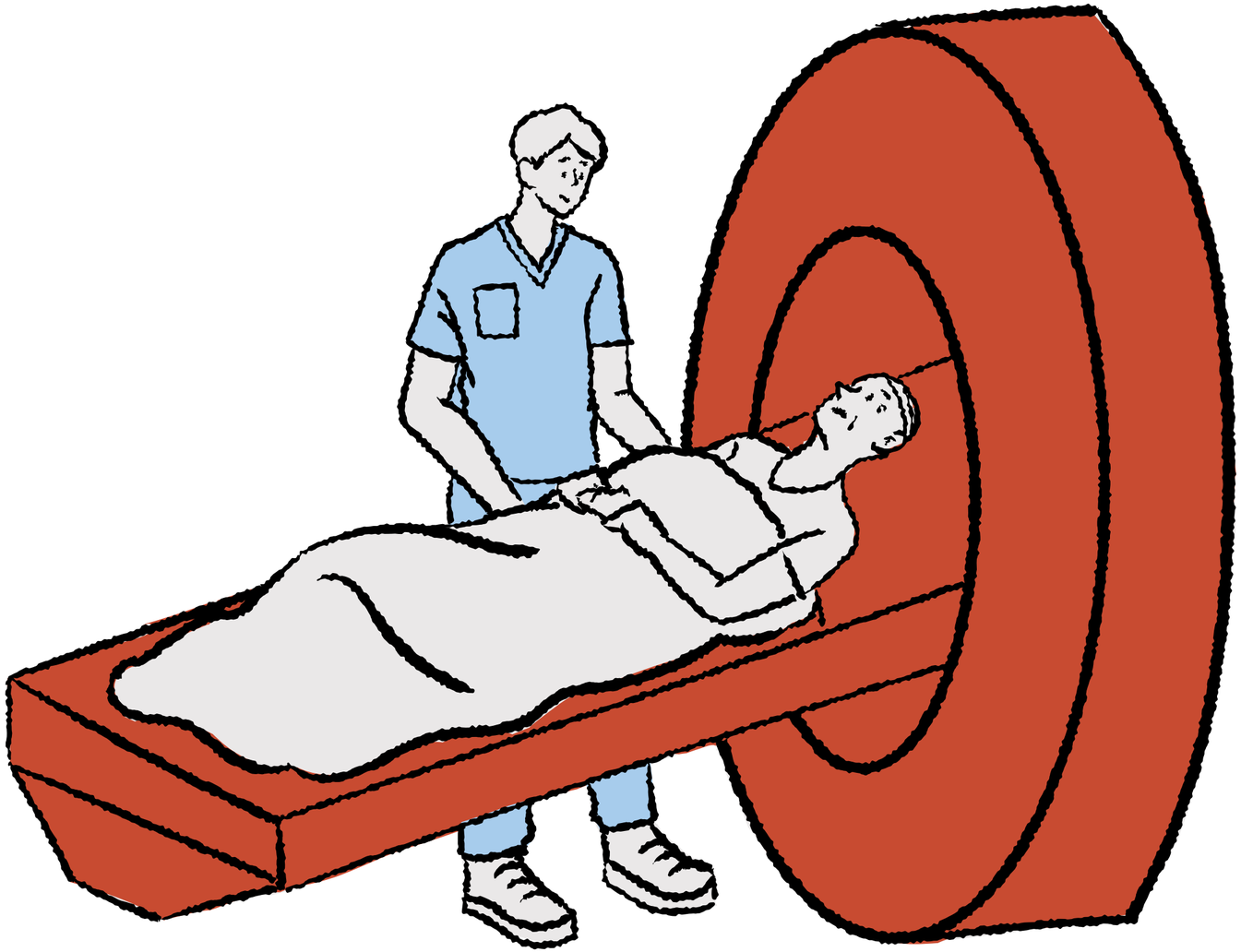
22q11 deletion



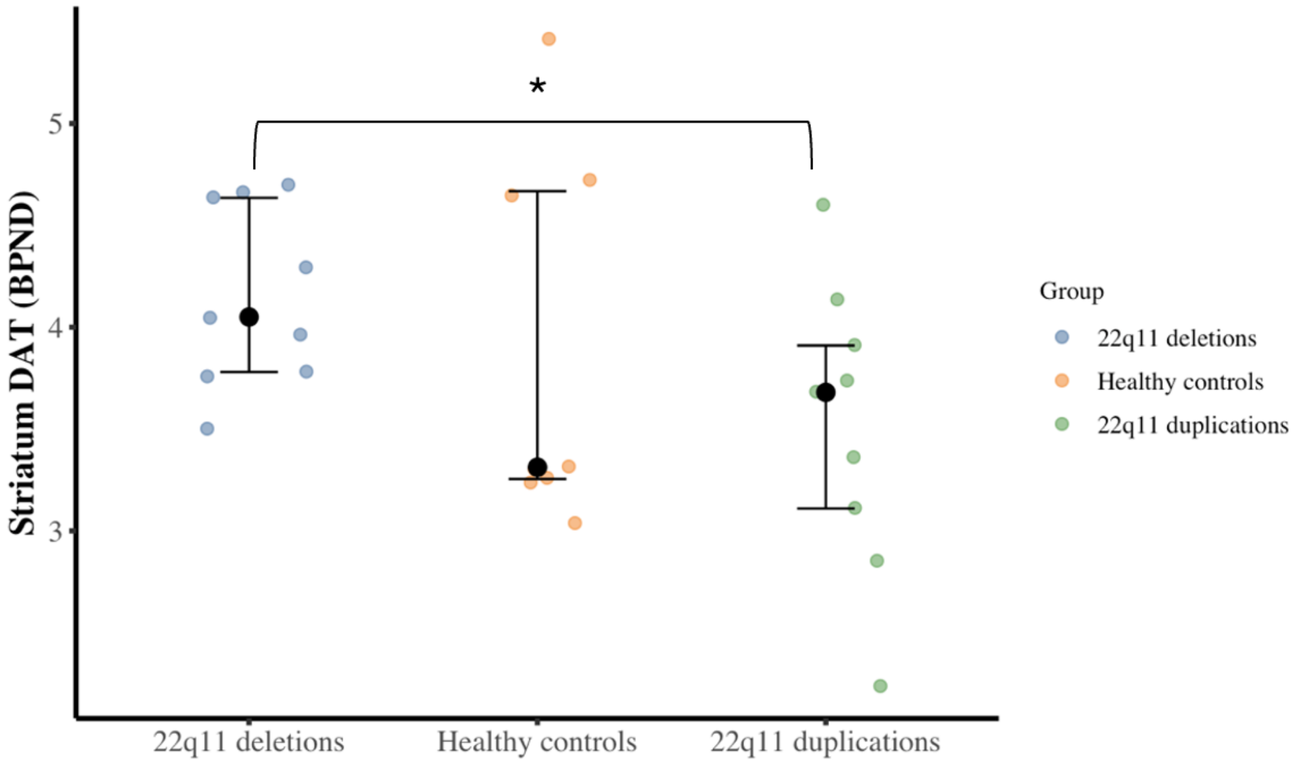
22q11 duplication

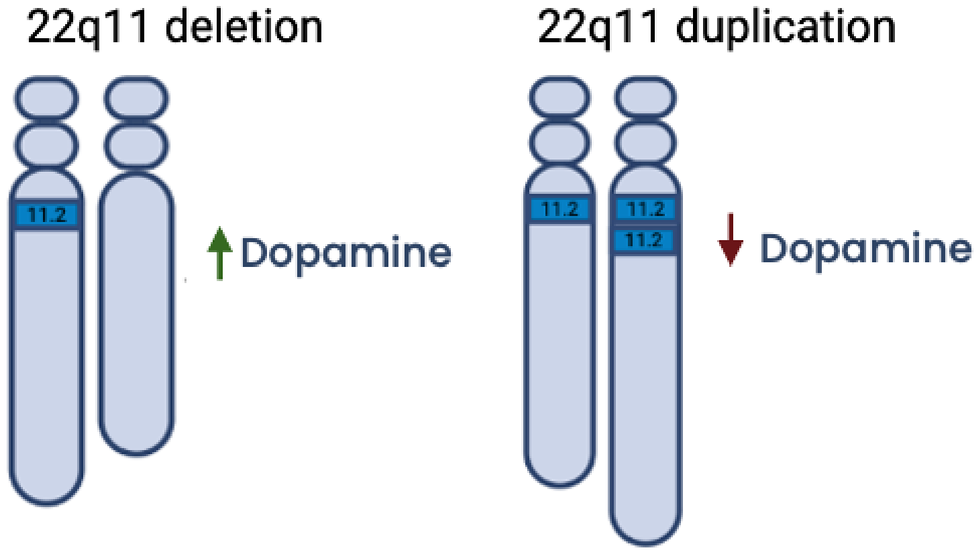


SPECT scan

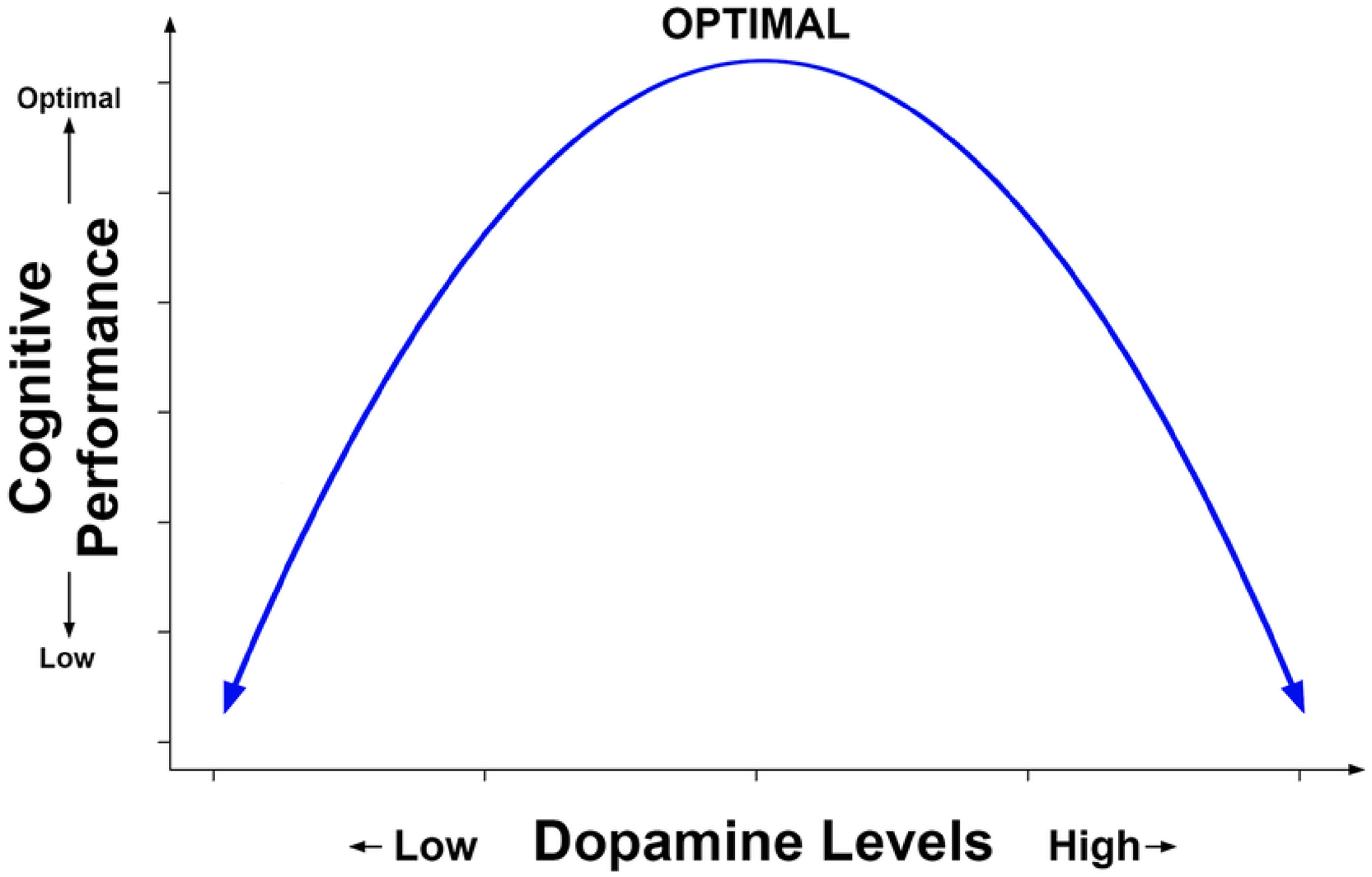


Results

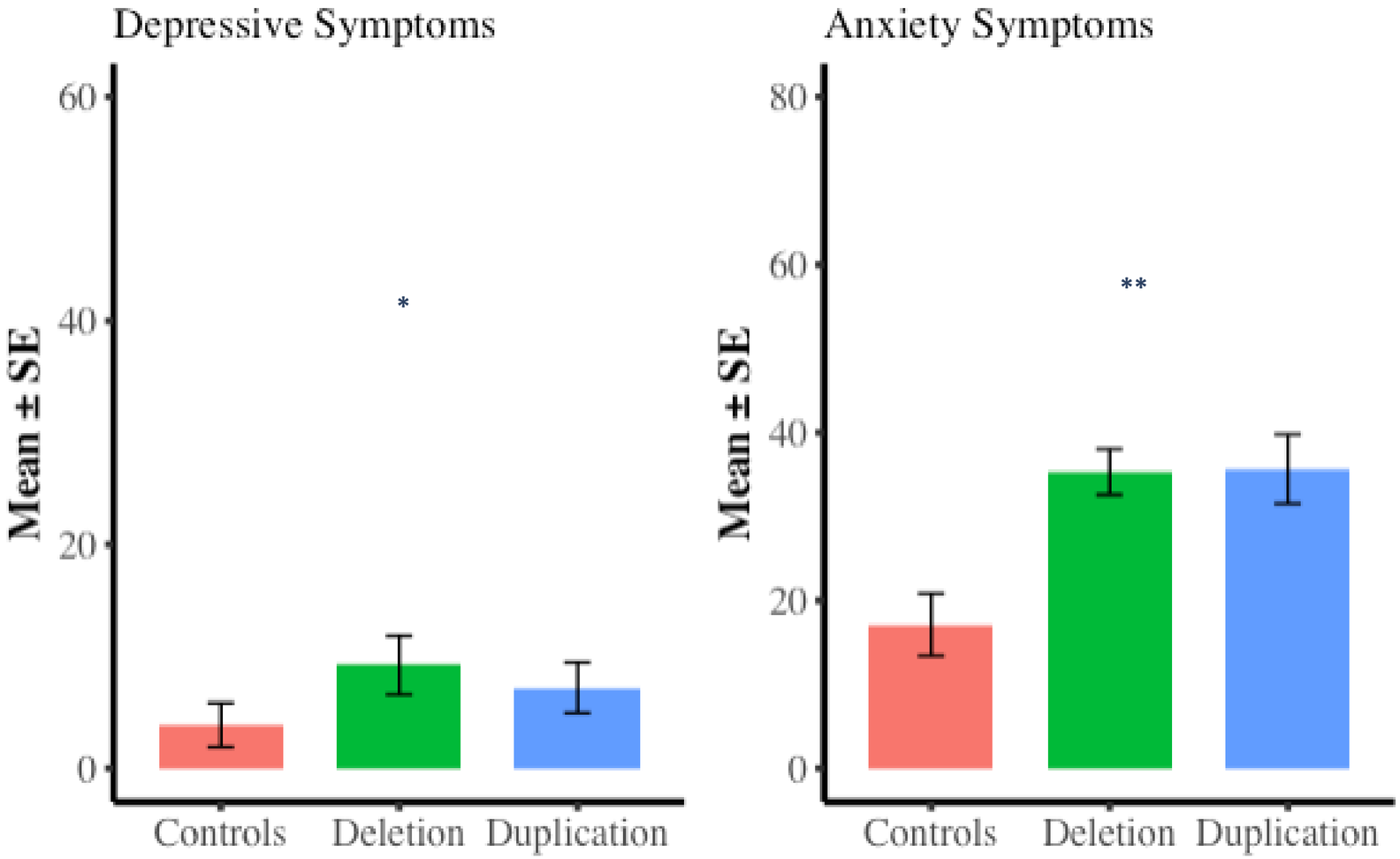

Striatum DAT (BPND)
5
4
3
*
22q11 deletions
Healthy controls
22q11 duplications
Group
22q11 deletions
Healthy controls
22q11 duplications


22q11 deletion
11.2
Dopamine
22q11 duplication
11.2
11.2
11.2
Dopamine

Inverted U-shape



Methylphenidate Treatment of Cognitive Dysfunction in Adults After Mild to Moderate Traumatic Brain Injury: Rationale, Efficacy, and Neural Mechanisms - Scientific Figure on ResearchGate. Available from: https://www.researchgate.net/figure/Representation-of-the-inverted-U-shaped-relation-of-prefrontal-dopamine-level-to_fig2_335760371 [accessed 21 Nov 2025]


Depressive Symptoms
Mean ± SE
60
40
20
0
*
Controls
Deletion
Duplication
Anxiety Symptoms
Mean ± SE
80
60
40
20
0
**
Controls
Deletion
Duplication

Closing remarks

- Deletions and duplications have similar problems.
- Deletions and duplications are biologically different.
- Should treatment and support be different?

Thank you for your attention any questions?

Researchteam

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- Sophie Kappert

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References

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