

Region-specific quantitative brain volumetry in fetuses with trisomy 21 by prenatal MRI

Eva Karner¹, Marlene Stuempflen², Michael Weber², Julia Binder¹, Christina Haberl¹, Sarah Glatter³, Patric Kienast², Daniela Prayer², Gregor Kasprian²

¹Department of Obstetrics and Feto-Maternal Medicine, Medical University of Vienna, Austria
²Department of Radiology, Division of Neuro- and Musculoskeletal Radiology, Medical University of Vienna, Austria
³Department of Pediatrics, Division of Neonatology, Medical University of Vienna, Austria

Objective

To characterize region-specific volumetric alterations in the fetal brain associated with Down Syndrome using prenatal MRI

Study metrics

- ✓ Retrospective analysis
- ✓ Single tertiary center for prenatal medicine
- ✓ Diagnosis of fetal Trisomy 21 + prenatal MRI

Methods

Fetal brain MRI of 76 pregnancies were performed and volumetric measurements of brain regions were conducted. The volumetric analysis of 38 fetuses with DS were compared to age-matched 38 neurotypical controls. Case/control volume ratios were calculated to obtain the percentage of reduction/increase in region specific brain volume.

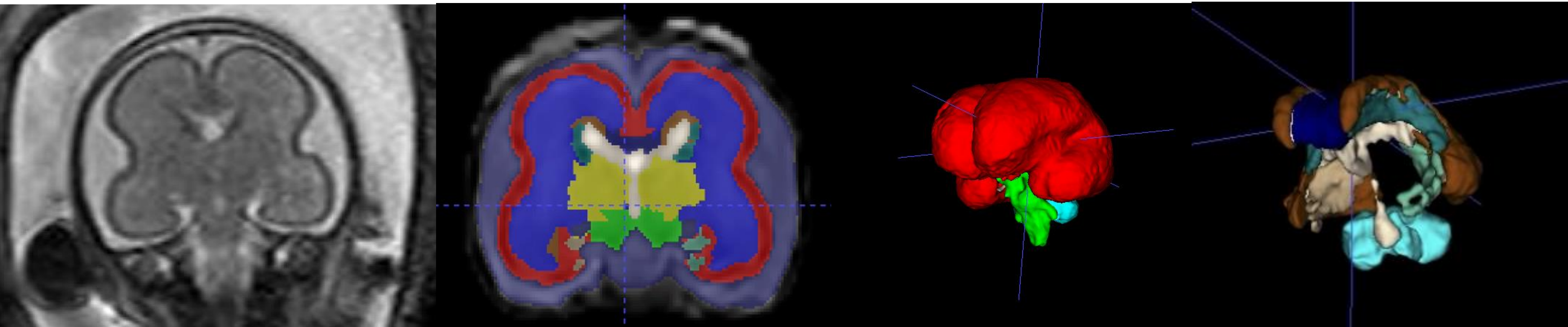


Figure 1 left to right: coronal plane of T2-weighted sequence with fetal MRI of a fetus with Down Syndrome in gestational week 27+0; superresolved reconstruction with ITK-snap with semi-augmented segmentation and manual correction; 3D reconstruction of cortex (red), brainstem (green) and cerebellum (turquoise); 3D reconstruction of ventricles (white), periventricular zone (brown), corpus callosum (blue), ganglionic eminence (dark green), hippocampi (beige and light green), cerebellum (turquoise)

Conclusion

This work describes significant region specific volume alterations in fetuses with DS by prenatal MRI. Most volume reduction was found in the Hippocampi, a region that is critically involved in cognition and memory. This suggests that structural changes underlying the neurological phenotype of DS emerge during the fetal period.

Fetal brain volume reduction

- Hippocampi 52% ↓
- Subcortical parenchyma 17% ↓
- Deep grey nuclei 16% ↓
- Corpus callosum 15% ↓
- Cerebellum 12% ↓
- Ganglionic eminence 12% ↓
- Total Brain Volume 5% ↓

Results

A significant decrease of fetal brain volume was observed in seven out of eleven analyzed regions as listed above. Total brain volume was 5% smaller in DS fetuses, without statistical significance ($p = 0.135$), indicating a region-specific pattern of brain volume reduction. Brain volumes between fetuses with congenital heart defects and those without did not show significant difference.

Volumetric differences in the ganglionic eminence increased with gestational age, indicating an early developmental alteration

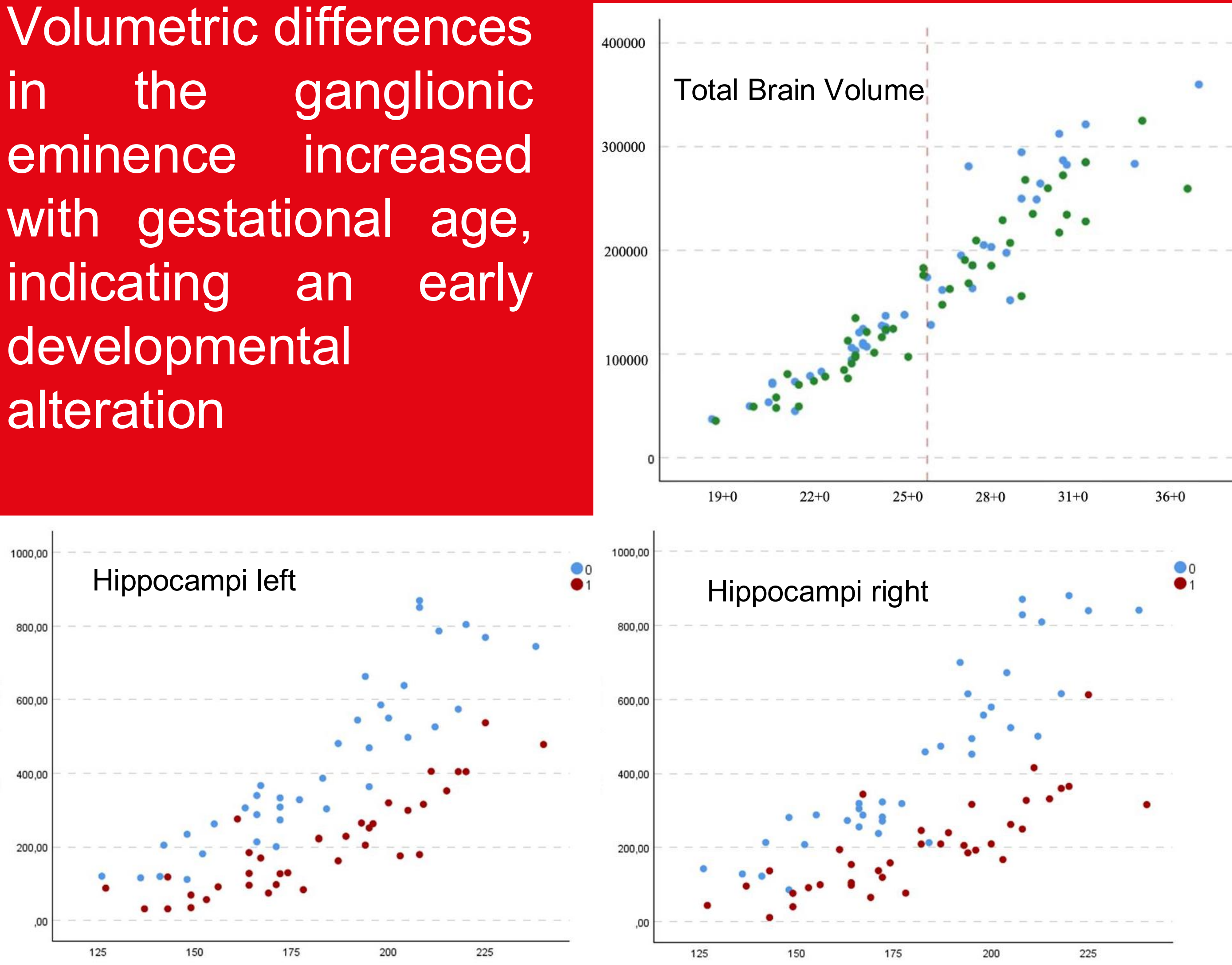


Figure 2 above: correlation of total brain volume and GA between cases and controls; left to right: volume of hippocampi and GA