

Voxel based morphometry in patients with albinism

B. Schmitz¹, G. Groen², T. N. Schaefer³, C. M. Krick⁴, B. Kaesmann-Kellner⁵

¹Dept. of Diagnostic Radiology, University Hospitals Ulm, Ulm, Germany, ²Dept. of Psychiatry, University Hospitals Ulm, Ulm, Germany,

³Neuroradiology, Saarland University Hospitals, Homburg, 66421, Germany, ⁴Neuroradiology, Saarland University Hospitals, Homburg, Germany, ⁵Ophthalmology, Saarland University Hospitals, Homburg, Germany

Introduction

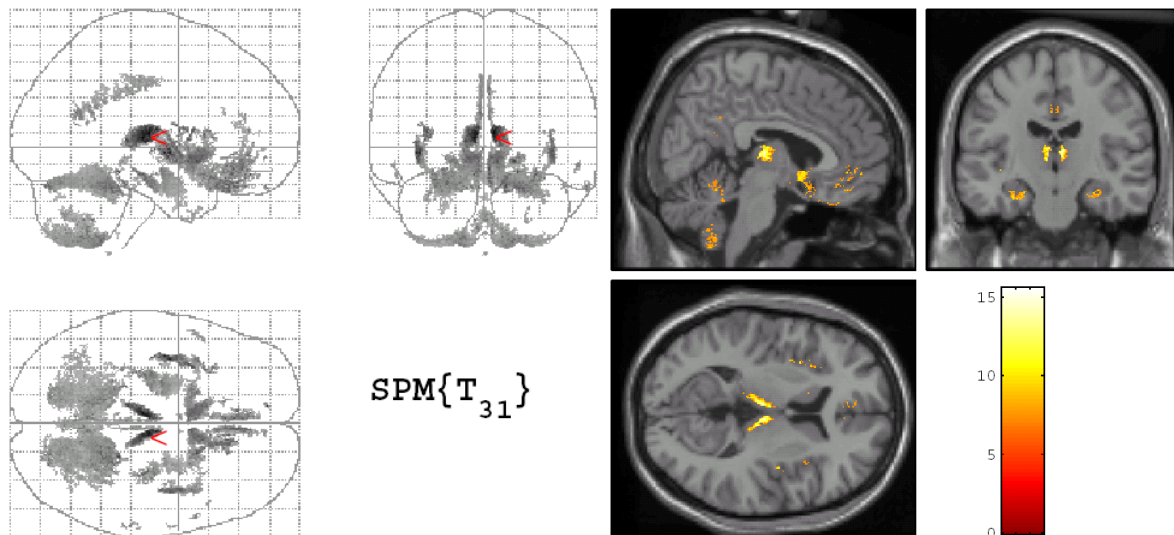
Patients with albinism are known to have many visual abnormalities including misrouting of optic fibers, low visual acuity and nystagmus. The functional architecture of primary visual cortex as well as the lateral geniculate nucleus is changed. However, it is not known whether there are morphologic differences in the brain of patients with albinism when compared to healthy controls.

Patients and Methods

17 patients with albinism diagnosed by a standardised clinical protocol as well as genetical testing and 15 age- and gender-matched normal controls underwent magnetic resonance imaging. Images were acquired (1.0 Tesla Magnetom Expert, Siemens, Germany) using an MPRAGE sequence with the following parameters: TR 15 ms, TE 7 ms, 1x1x1 mm isotropic voxels. Regional differences in gray matter density were examined by voxel based morphometry using SPM2 (Statistical Parametric Mapping, Wellcome Department of Cognitive Neurology, London). Group comparisons were calculated by means of two-sample t-tests ($p < 0.05$, corrected for multiple comparisons).

Results

Bilateral between-group gray matter differences were highly symmetric and showed regionally decreased gray matter density in patients with albinism in several regions: pulvinar, nucleus accumbens, anterior caudate, putamen, anterior insula, head of the hippocampus, amygdala (Figures). To a lesser extent, grey matter densities were also reduced in the cerebellum and anterior and posterior parts of cingulate gyrus.



Discussion

Reductions of gray matter densities in patients with albinism comprised several cortical and subcortical structures. These may reflect adaptive changes to abnormalities of the visual system, but may also be caused by the genetic abnormality. Histologically known changes of the primary visual cortex remarkably do not correlate with reduction of gray matter density in these areas.