FRACTAL COMPLEXITY AND THICKNESS OF THE HUMAN CORTEX IS INCREASED IN WILLIAMS SYNDROME

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We report the first 3D brain maps to reveal the spatial pattern of cortical abnormalities in Williams syndrome (WS), a genetic condition associated with deletion of ~20 contiguous genes on chromosome 7. We mapped a striking pattern of abnormally increased cortical thickness and gyral complexity in WS, using new algorithms to measure the fractal dimension of the human cerebral cortex.

Methods. 75 T1-weighted 3D MRI brain scans (slice thickness 1.2 mm; 1.5 Tesla) were acquired from 36 subjects with genetically-confirmed Williams syndrome (mean age: 29.3±1.6SE years, 14M/22F) and 39 age-matched healthy controls (age: 29.3±1.6 years; 16M/23F). 72 sulcal landmarks per brain were traced on 3D cortical surface models extracted from each scan. The rate at which cortical surface area increased with increasing spatial frequency was used to measure the fractal complexity of the cortex. The profile of cortical thickness was also computed across the entire cortex by applying the Eikonal fire equation to individual 3D maps of gray matter distribution. Cortical thickness profiles were averaged and compared across groups.

Findings. Left and right hemisphere complexity in the Williams group was greater than that in controls (L hem.: 2.2509±0.0018SE v. 2.2450±0.0017; p<0.019; R hem.: 2.2482±0.0018 v. 2.2413±0.0015; p<0.0019). WS subjects had greater cortical thickness (by approx. 10%; p<0.05) in a broad contiguous region including perisylvian language cortices bilaterally, but had thinner cortices in frontal-parietal and cingulate regions (p<0.05). These results visualize the profile of cortical abnormalities suggested by prior reports of gyral pattern anomalies and polymicrogyria in Williams syndrome. Disturbed developmental processes during cortical pattern formation and gyrogenesis may increase cortical complexity and thickness.