



Congenital unilateral upper limb absence flattens the contralateral hand knob

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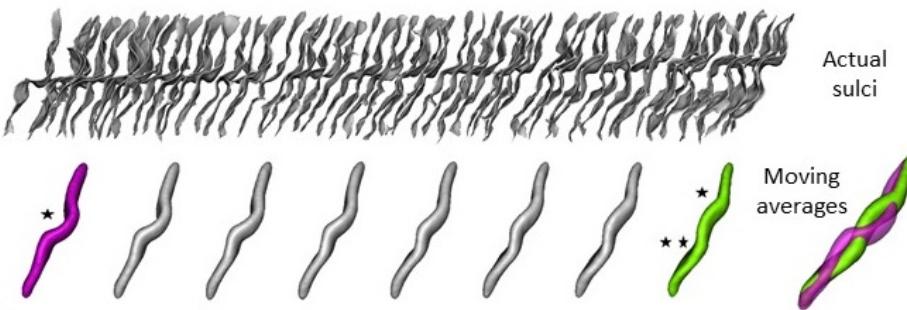
Introduction:

We are seven billion humans with unique fingerprints but also unique cortical folding patterns. The cortical folding process occurs essentially during the last trimester of pregnancy. The forces driving the folding dynamics are still unclear but several hypotheses assume interactions with the primal sketch of the cortical architecture [1]. Hence, the folding patterns could be a useful proxy of the variability of the cortical architecture and may embed the signature of abnormal developmental events associated for instance to cognitive efficacy [2] or psychiatric symptoms [3]. In this abstract, we explore this concept in the context of congenital unilateral upper limb absence [4], hereafter termed congenital one-handers. To test whether the folding pattern signatures can be specific to developmental events, we compare congenital one-handers with individuals with acquired unilateral upper limb amputation [5] and controls.

Methods:

This study relies on T1-weighted MPRAGE images collected using 3T Siemens scanners (Vario and Prisma) with 1mm isotropic resolution. 16 congenital one-handers (four with absent right hand), 16 amputees (four with absent right hand) and 24 two-handed controls were recruited. We focus on the shape of the central sulcus, using an approach developed previously to study morphological correlates of handedness [8]. First, the two central sulci were extracted automatically using the "Morphologist 2012" toolbox of BrainVisa [<http://brainvisa.info>] [9,10] and brought to Talairach space with an affine transform. Right sulci were flipped to become comparable with left sulci. Then, the shape similarity between two sulci was coded by the average quadratic distance between their 3D representations after pairwise rigid alignment. Finally, from the resulting similarity matrix, we used Isomap-based manifold learning to capture a bidimensional approximation of the high dimensional space spanned by each sulcus. For each Isomap dimension, we finally computed local average sulci at regularly spaced locations to clarify visually the shape variations encoded along the manifold. Hence, localization of a specific sulcus in the manifold provides a summary of its shape. We compared the localizations of the central sulci of the three groups using two-sided Wilcoxon tests.

A: First dimension of the Central Sulcus manifold in the general population



B: Central sulcus manifold in a population gathering one-handers and two-handed controls

First dimension:



Distribution of actual sulci:



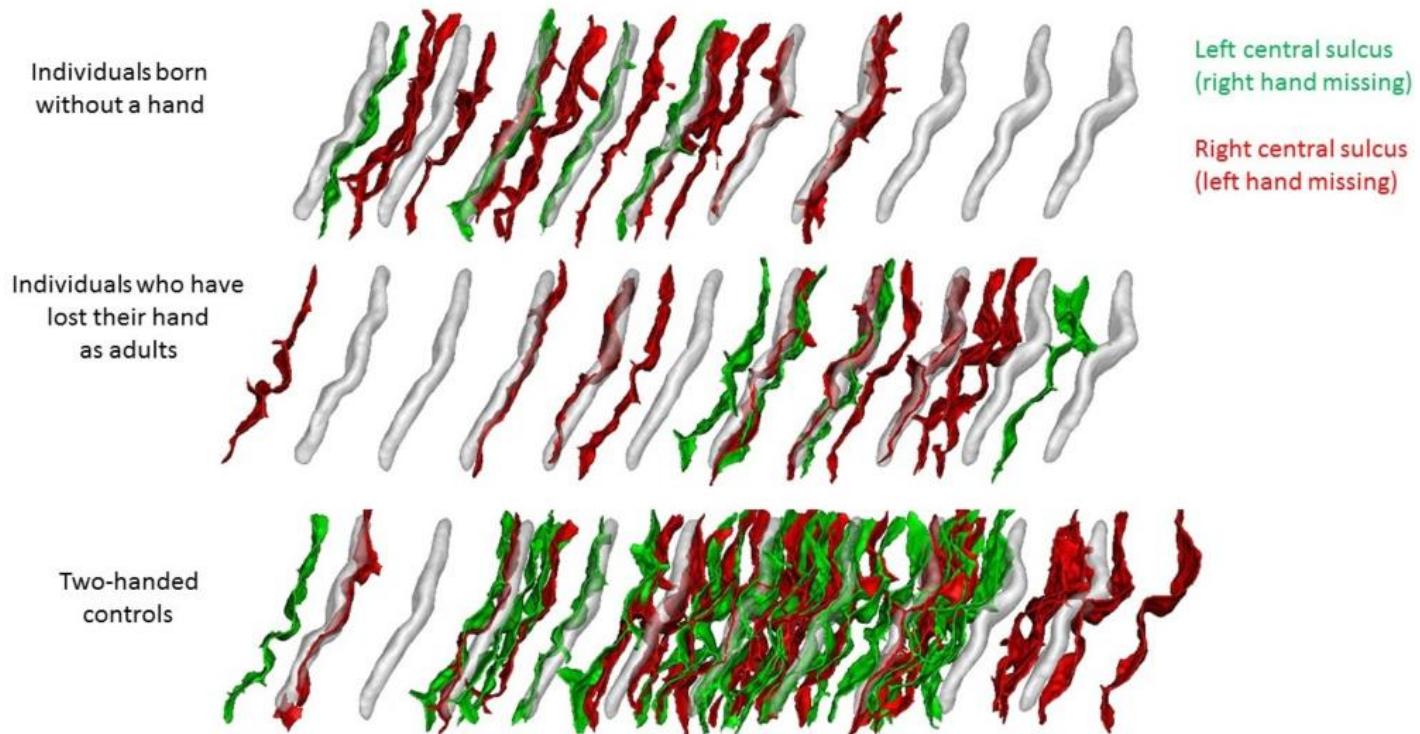
Second dimension:



Results:

In the past, we have been computing the shape-manifold of the central sulcus for over ten different databases. In each study, the first dimension of the manifold, namely the shape variations accounting for the largest amount of variance, did encode the transition from a single knob configuration toward a two knobs configuration [7,8] (see Fig 1.A). In contrast, in our study of one-handers, this specific shape variation is encoded by the second dimension of the Isomap only (see Fig 1.B). The first dimension encodes the transition from a flat sulcus toward a sulcus with a marked hand knob. This means that a large proportion of one-handers have at least one flat sulcus, which is an unusual configuration in the general population. For congenital one-handers, we found that the sulci contralateral to the missing hand are much more flattened than the two sulci of the controls ($p=0.0002$), which is not the case for their ipsilateral sulci ($p>0.5$) (Fig. 2). We did not find any difference between amputees and controls.

Shape of the Central Sulcus contralateral to the missing hand of one-handers



Conclusions:

Congenital hand deformities usually occur between the fourth and the eighth week after the embryo is formed [6]. Our study has shown that the aftermath regarding brain development prints a signature in the cortical folding morphology, a flattened hand knob. Furthermore, the absence of this signature after amputation (more than 10 years after for some amputees), shows that folding patterns are stable enough to encode developmental signatures resistant to adverse events occurring later in life. Future studies will tell us whether the extent of the remnant hand knob in the congenital one-handers depends on their usage of the handless arm in daily tasks.

Imaging Methods:

Anatomical MRI

Modeling and Analysis Methods:

Image Registration and Computational Anatomy ²

Neuroanatomy:

Cortical Anatomy and Brain Mapping ¹

Keywords:

Computational Neuroscience
Machine Learning
Morphometrics
Motor
Spatial Normalization
STRUCTURAL MRI

^{1|2}Indicates the priority used for review

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Please indicate which methods were used in your research:

Structural MRI

For human MRI, what field strength scanner do you use?

3.0T

Provide references in author date format

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