

NeuroPsicologia dello
Sviluppo e
Riabilitazione

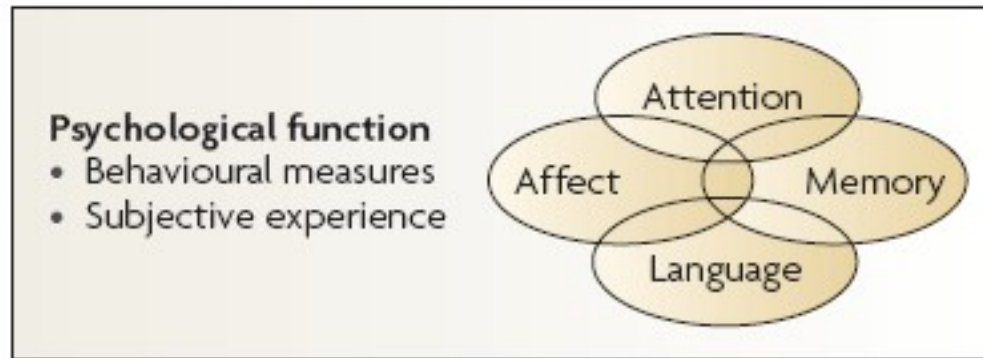
Padova, 21 Ottobre 2020

Lezione n 6

Dalla Genetica al comportamento???

b

Intermediate-phenotype approach (cognitive neurogenetics)



Behavioural genetics

Cognitive neuroscience

Genetic variation

- Single polymorphic loci
- Functional interactions among loci (genetic networks)
- Epigenetic factors



Brain-based intermediate phenotype

- Localized activation
- Functional connectivity (brain networks)
- Volume and structure
- Evoked electrical activity
- Neurochemical assays





DIAGNOSTIC AND STATISTICAL
MANUAL OF
MENTAL DISORDERS

FIFTH EDITION

DSM-5

AMERICAN PSYCHIATRIC ASSOCIATION

Specific Learning Disorder (66)

___.__ (___.__)

Specific Learning Disorder (66)

Specify if:

315.00 (F81.0)

With impairment in reading (*specify if with word reading accuracy, reading rate or fluency, reading comprehension*)

315.2 (F81.81)

With impairment in written expression (*specify if with spelling accuracy, grammar and punctuation accuracy, clarity or organization of written expression*)

315.1 (F81.2)

With impairment in mathematics (*specify if with number sense, memorization of arithmetic facts, accurate or fluent calculation, accurate math reasoning*)

Specify current severity: Mild, Moderate, Severe

Prevalence

The prevalence of specific learning disorder across the academic domains of reading, writing, and mathematics is 5%–15% among school-age children across different languages and cultures. Prevalence in adults is unknown but appears to be approximately 4%.

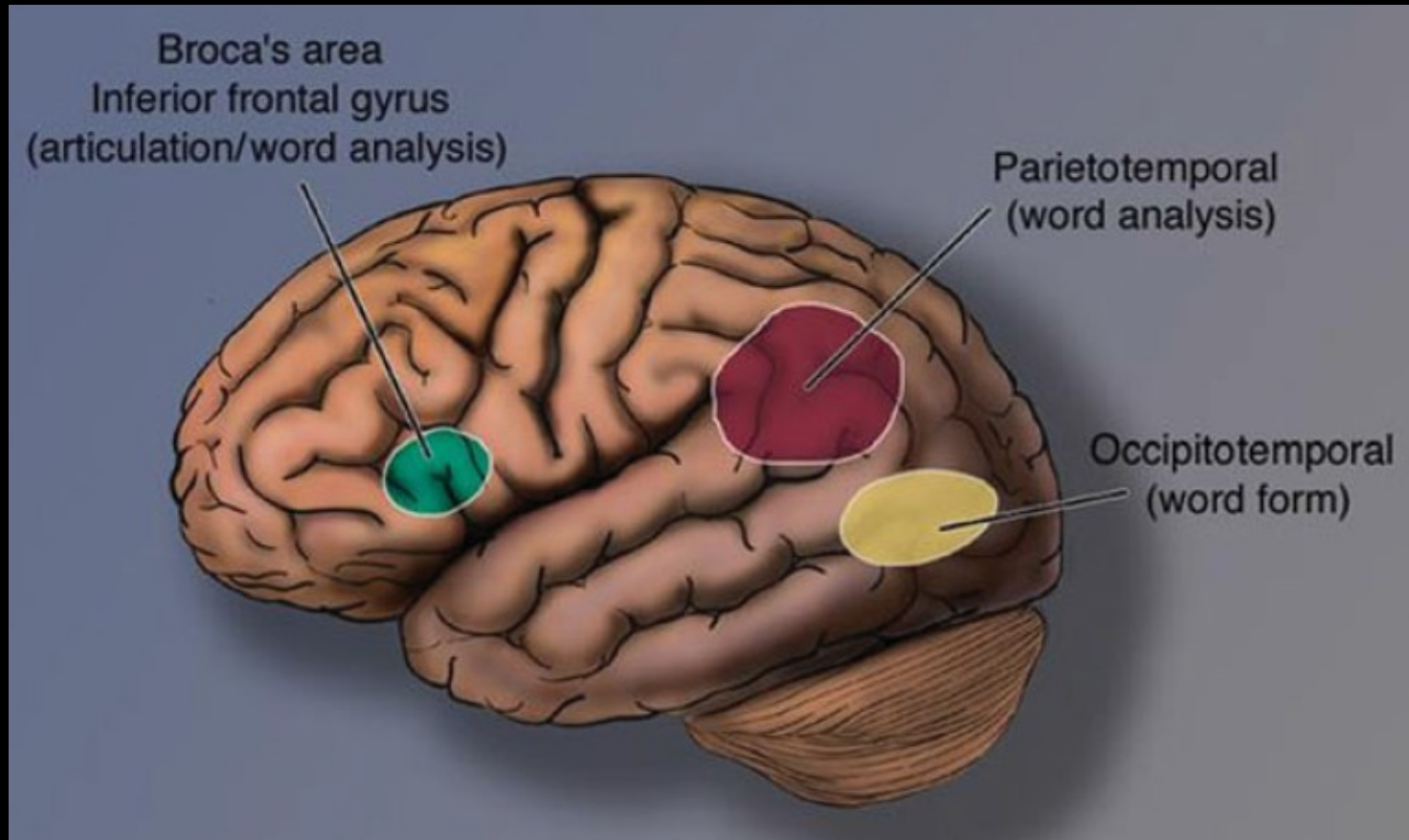
Gender-Related Diagnostic Issues

Specific learning disorder is more common in males than in females (ratios range from about 2:1 to 3:1) and cannot be attributed to factors such as ascertainment bias, definitional or measurement variation, language, race, or socioeconomic status.

Comorbidity

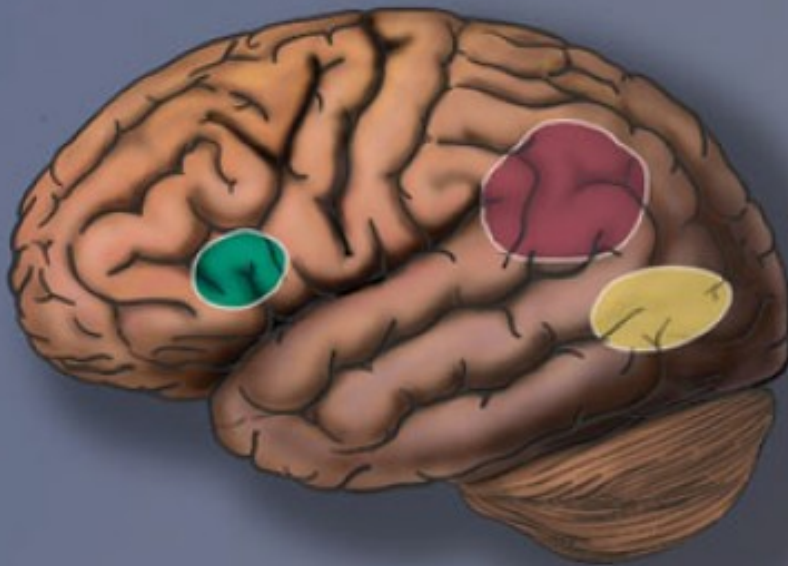
Specific learning disorder commonly co-occurs with neurodevelopmental (e.g., ADHD, communication disorders, developmental coordination disorder, autistic spectrum disorder) or other mental disorders (e.g., anxiety disorders, depressive and bipolar disorders). These comorbidities do not necessarily exclude the diagnosis specific learning disorder but may make testing and differential diagnosis more difficult, because each of the co-occurring disorders independently interferes with the execution of activities of daily living, including learning. Thus, clinical judgment is required to attribute such impairment to learning difficulties. If there is an indication that another diagnosis could account for the difficulties learning keystone academic skills described in Criterion A, specific learning disorder should not be diagnosed.

La neurobiologia della lettura

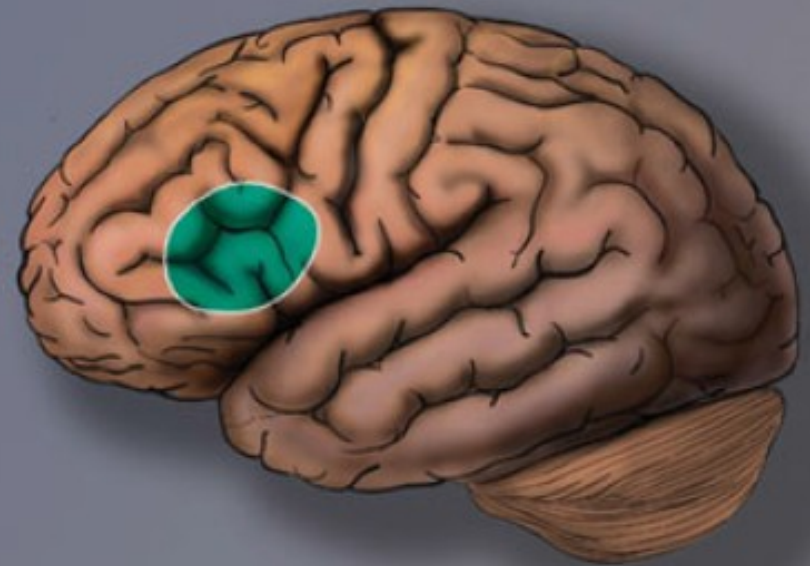


Un ringraziamento speciale a Kenneth Pugh (e Principe Marconi)

I disordini fonologici e ortografici: Cause or Effetti della dislessia?

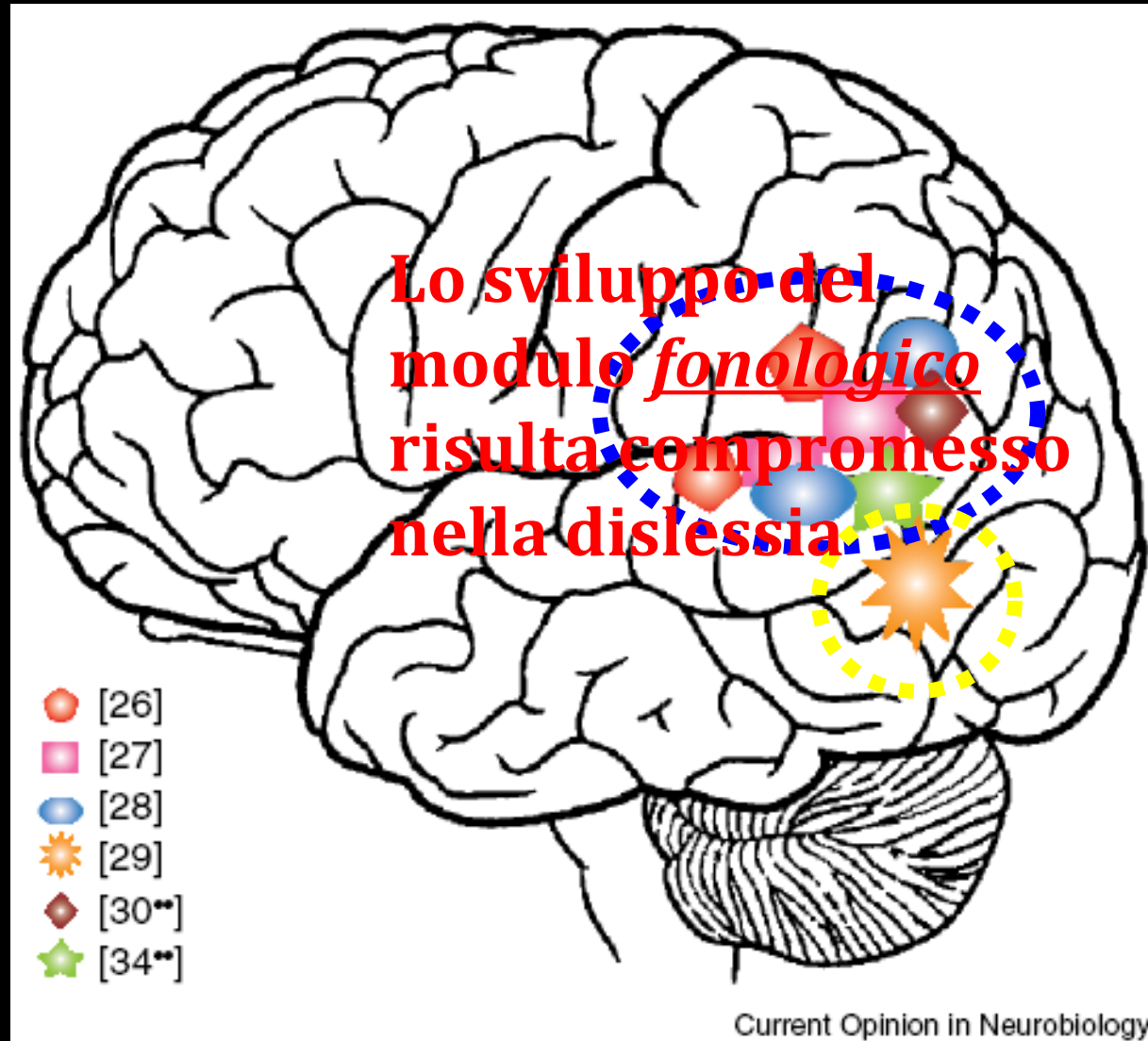


Nonimpaired



Dyslexic

Sono veramente queste le basi neurobiologiche della dislessia???



From Temple (2002)

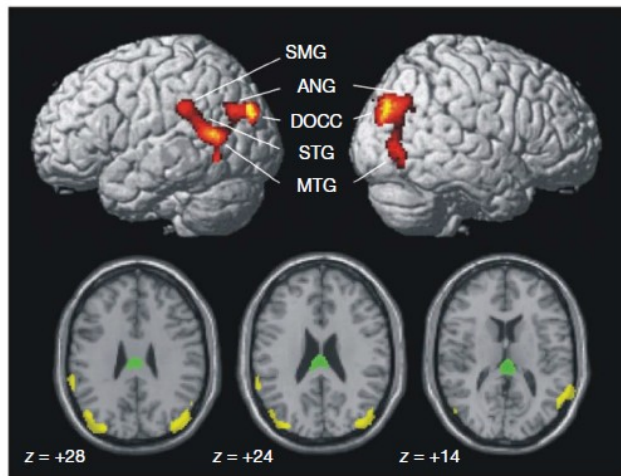
Il deficit Fonologico: Causa o Effetto della Dislessia?

Late-literates had more white matter in the splenium of the corpus callosum and more grey matter in bilateral angular, dorsal occipital, middle temporal, left supramarginal and superior temporal gyri. The importance of these brain regions for skilled reading was investigated in early literates, who learnt to read as children. We

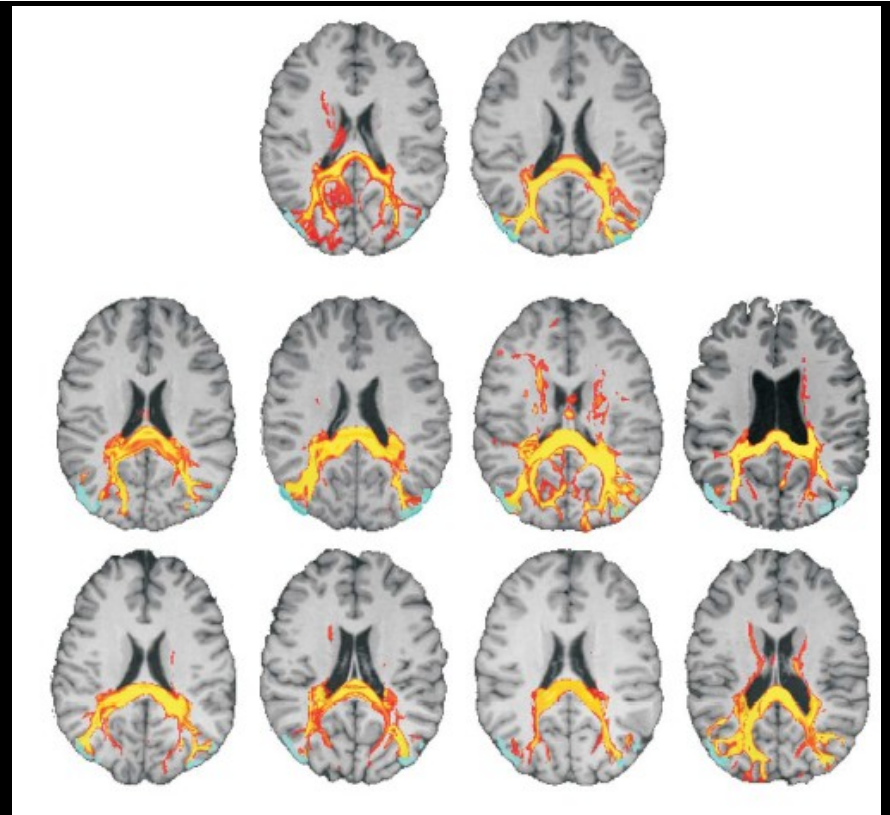
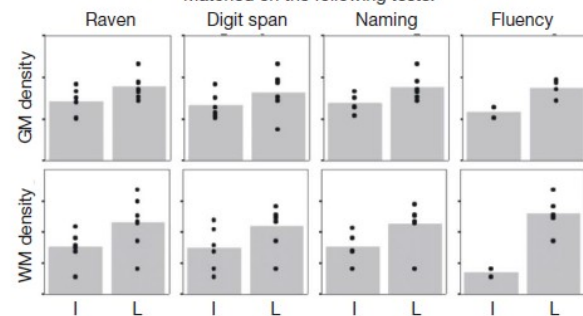
LETTERS

An anatomical signature for literacy

Manuel Carreiras^{1,2,3,4}, Mohamed L. Seghier⁵, Silvia Baquero⁶, Adelina Estévez⁴, Alfonso Lozano⁶, Joseph T. Devlin⁷ & Cathy J. Price⁵



Matched on the following tests:



OPINION

Illiterate to literate: behavioural and cerebral changes induced by reading acquisition

Stanislas Dehaene, Laurent Cohen, José Morais and Régine Kolinsky

Abstract | The acquisition of literacy transforms the human brain. By reviewing studies of illiterate subjects, we propose specific hypotheses on how the functions of core brain systems are partially reoriented or ‘recycled’ when learning to read. Literacy acquisition improves early visual processing and reorganizes the ventral occipito-temporal pathway: responses to written characters are increased in the left occipito-temporal sulcus, whereas responses to faces shift towards the right hemisphere. Literacy also modifies phonological coding and strengthens the functional and anatomical link between phonemic and graphemic representations. Literacy acquisition therefore provides a remarkable example of how the brain reorganizes to accommodate a novel cultural skill.

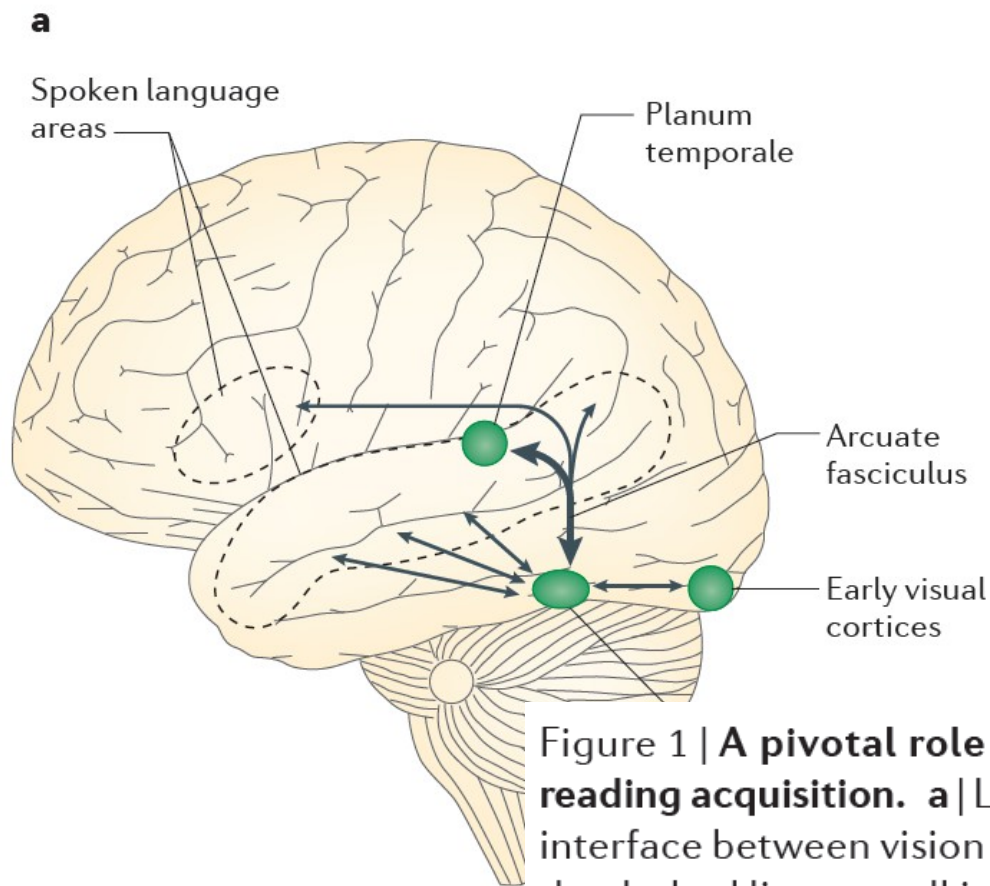
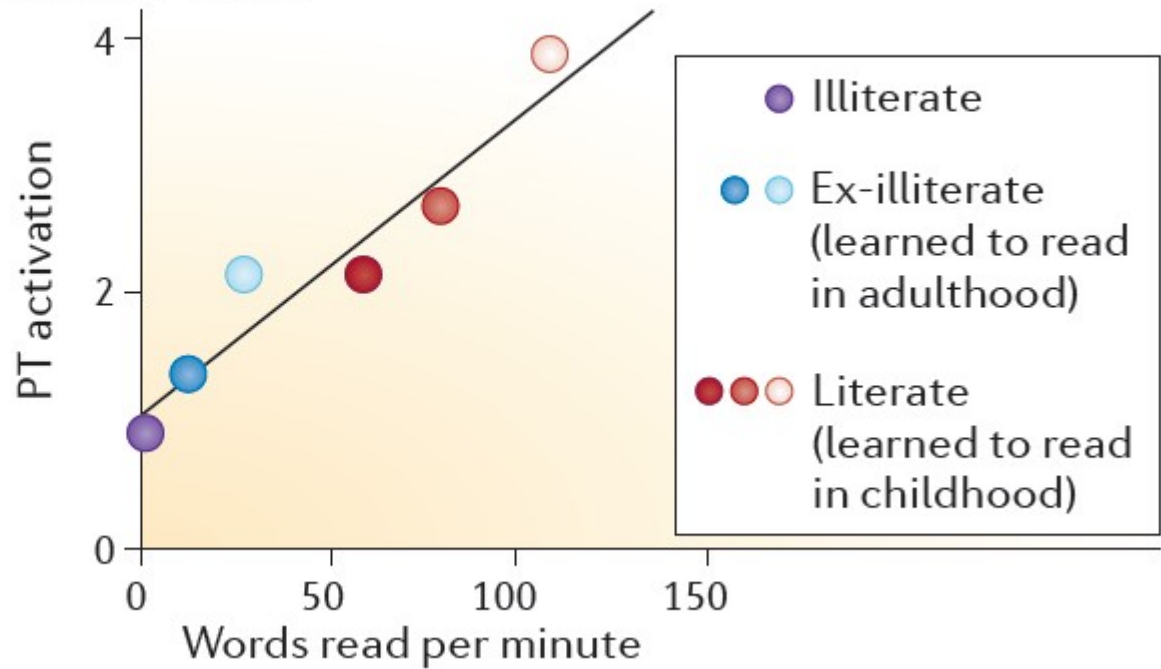
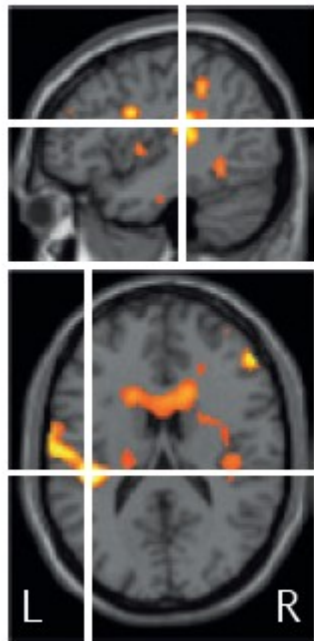
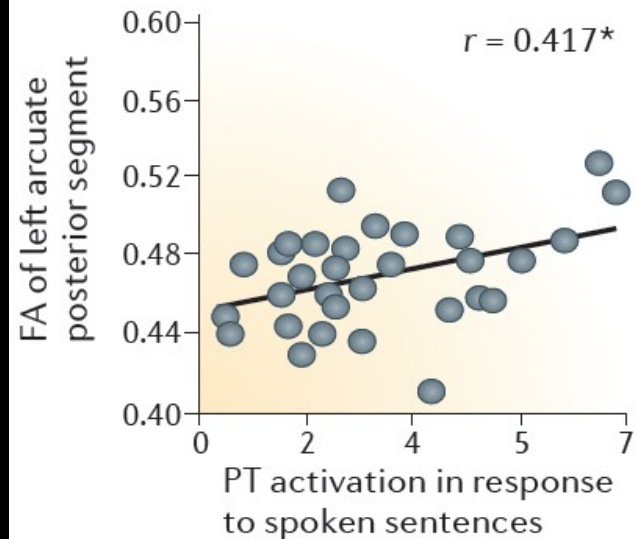
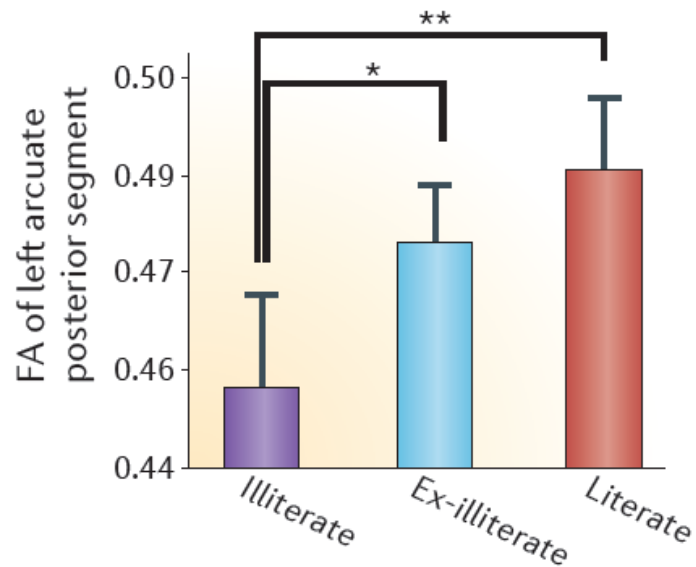


Figure 1 | A pivotal role of the ventral occipito-temporal cortex in reading acquisition. **a** | Learning to read involves developing an efficient interface between vision and spoken language. The regions outlined by the dashed lines are all involved in processing spoken language prior to reading acquisition^{95,178}. The visual word form area (VWFA), which is located in the ventral left occipito-temporal sulcus, and its afferents and efferents are thought to play a pivotal role in reading acquisition by enabling the rapid recognition of strings of letters and their translation into sequences of sounds. The regions in green — the VWFA, early visual cortices (V1 and V2) and planum temporale are all demonstrably enhanced by reading acquisition. The connections between them (shown by arrows) may also be enhanced — this has been directly demonstrated in the case of the posterior part of the arcuate fasciculus (thick arrow). **b–d** | The

a Increased response to spoken words



c Increased connectivity with the VWFA



b Letter-sound congruence

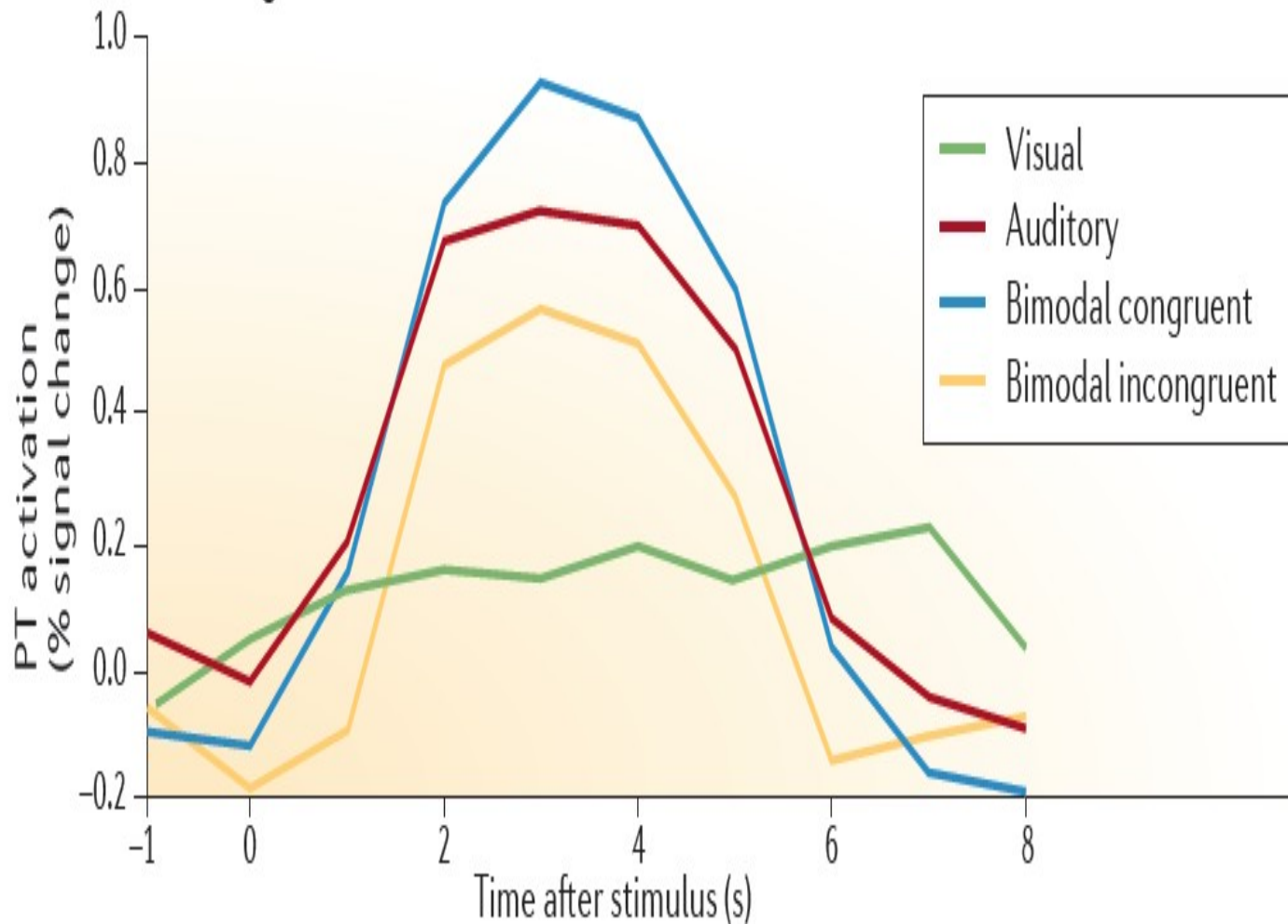


Figure 2 | **Impact of reading acquisition on the planum temporale.** Reading acquisition transforms the left planum temporale (PT) and the surrounding superior temporal cortex (a region involved in the auditory processing of speech). **a** | Enhanced brain responses to speech. Coloured

La Neuro-plasticità è alla base dello sviluppo cerebrale

How Learning to Read Changes the Cortical Networks for Vision and Language

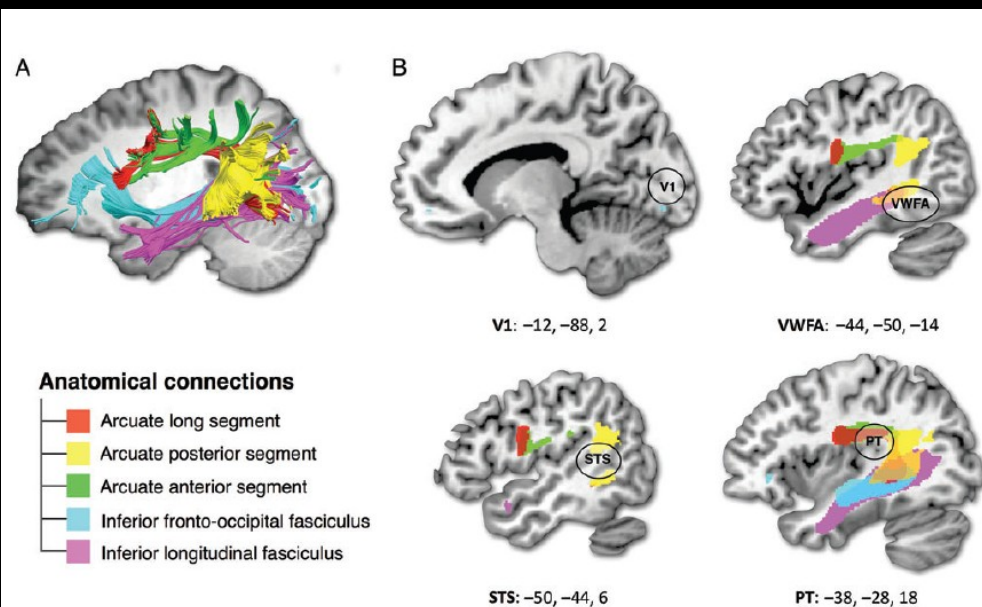
Stanislas Dehaene,^{1,2,3,4*} Felipe Pegado,^{1,2,3} Lucia W. Braga,⁵ Paulo Ventura,⁶ Gilberto Nunes Filho,⁵ Antoinette Jobert,^{1,2,3} Ghislaine Dehaene-Lambertz,^{1,2,3} Régine Kolinsky,^{7,8} José Morais,⁷ Laurent Cohen^{9,10,11}

Does literacy improve brain function? Does it also entail losses? Using functional magnetic resonance imaging, we measured brain responses to spoken and written language, visual faces, houses, tools, and checkers in adults of variable literacy (10 were illiterate, 22 became literate as adults, and 31 were literate in childhood). As literacy enhanced the left fusiform activation evoked by writing, it induced a small competition with faces at this location, but also broadly enhanced visual responses in fusiform and occipital cortex, extending to area V1. Literacy also enhanced phonological activation to speech in the planum temporale and afforded a top-down activation of orthography from spoken inputs. Most changes occurred even when literacy was acquired in adulthood, emphasizing that both childhood and adult education can profoundly refine cortical organization.

Learning to Read Improves the Structure of the Arcuate Fasciculus

Michel Thiebaut de Schotten^{1,2,3}, Laurent Cohen^{3,4,5}, Eduardo Amemiya⁶, Lucia W. Braga⁶ and Stanislas Dehaene^{7,8,9,10}

The acquisition of literacy results from an effortful learning process that leads to functional changes in several cortical regions. We explored whether learning to read also leads to anatomical changes within the left intrahemispheric white matter pathways that interconnect these regions. Using diffusion tensor imaging tractography, we compared illiterates with ex-illiterates who learned to read during adulthood and literates who learned to read during their childhood. Literacy related to an increase in fractional anisotropy and a decrease in perpendicular diffusivity in the temporo-parietal portion of the left arcuate fasciculus. The microstructure within this pathway correlated with the reading performance and the degree of functional activation within 2 dominant brain regions involved in reading: The Visual Word Form Area in response to letter strings, and the posterior superior temporal cortex in response to spoken language. Thus, the acquisition of literacy is associated with a reinforcement of left temporo-parietal connections whose microstructure predicts overall reading performance and the functional specialization of the Visual Word Form Area. This anatomical magnetic resonance imaging marker may be useful to predict developmental reading disorders.



Diagnosing the Neural Circuitry of Reading

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<http://dx.doi.org/10.1016/j.neuron.2017.08.007>

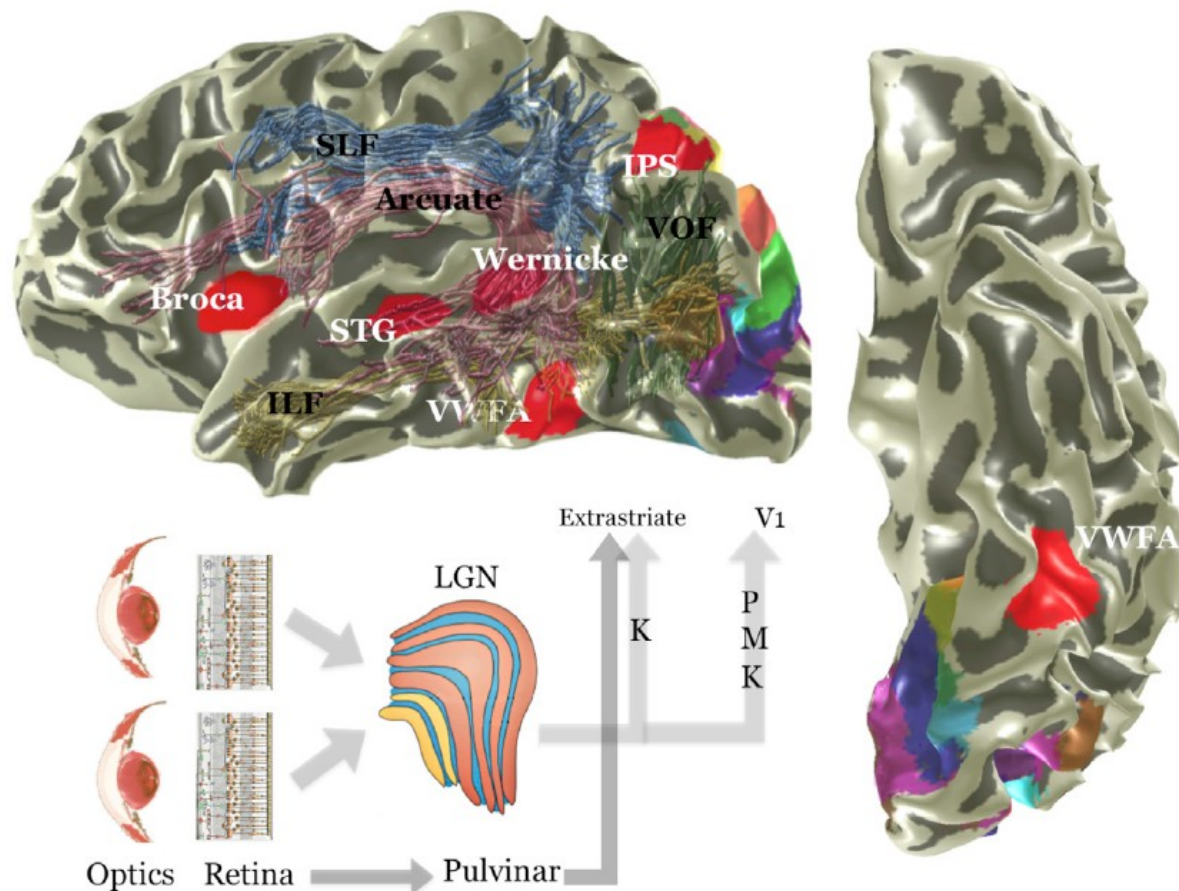


Figure 1. The Reading Circuitry

Retinal photoreceptors encode the image and then transform this encoding with multiple specialized neural circuits. Retinal ganglion cells project directly to the lateral geniculate nucleus (LGN) and via the superior colliculus to the pulvinar. The parvocellular (P), magnocellular (M), and koniocellular (K) layers of the LGN project to the V1 and extrastriate cortex. The visual regions within the pulvinar project mainly to the extrastriate cortex. These thalamic nuclei also receive many inputs from the cortex. The colors overlaid on the posterior cortex show the locations of some visual field maps in the posterior cortex. Learning to read produces specialized circuitry that selects certain signals for further processing. Posterior visual signals are transmitted to the auditory and language system (red overlay, white text). Five general cortical regions contain subdivisions that are consistently identified as active during reading: the VOT, which includes the visual word form area (VVFA); regions within the intraparietal sulcus (IPS), which appear to be a source of top-down modulation; regions near the primary auditory cortex in the superior temporal gyrus (STG), where adult dyslexics have low activation while integrating letters with speech sounds; and Wernicke's area and Broca's area, which are implicated in the comprehension and production of language.

Four large tracts (black text) terminate near the functionally defined regions and/or contain subdivisions with tissue properties that are consistently found to differ between groups of good and poor readers: the VOF, ILF, superior longitudinal fasciculus (SLF), and the AF (Arcuate).

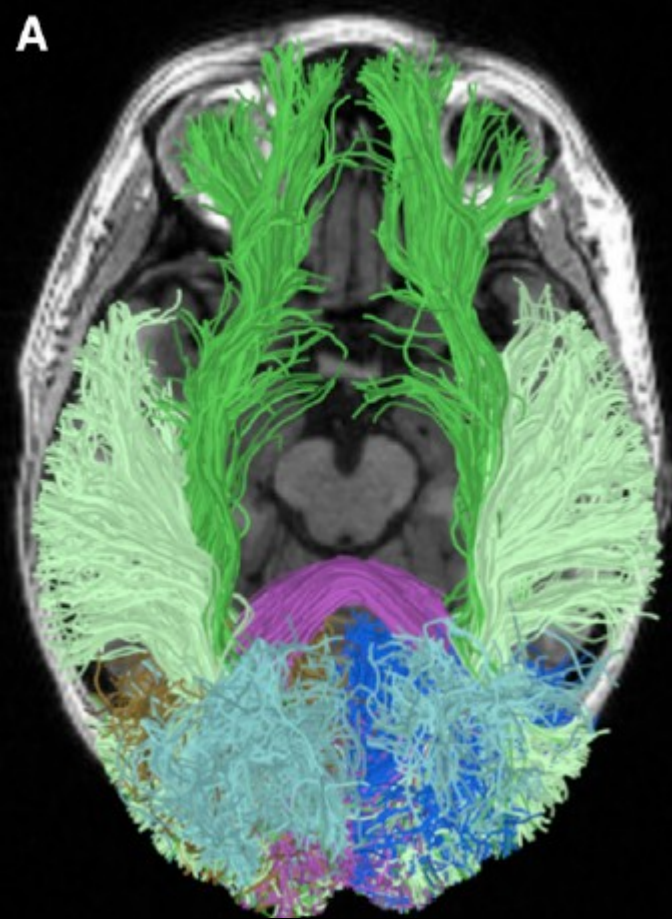
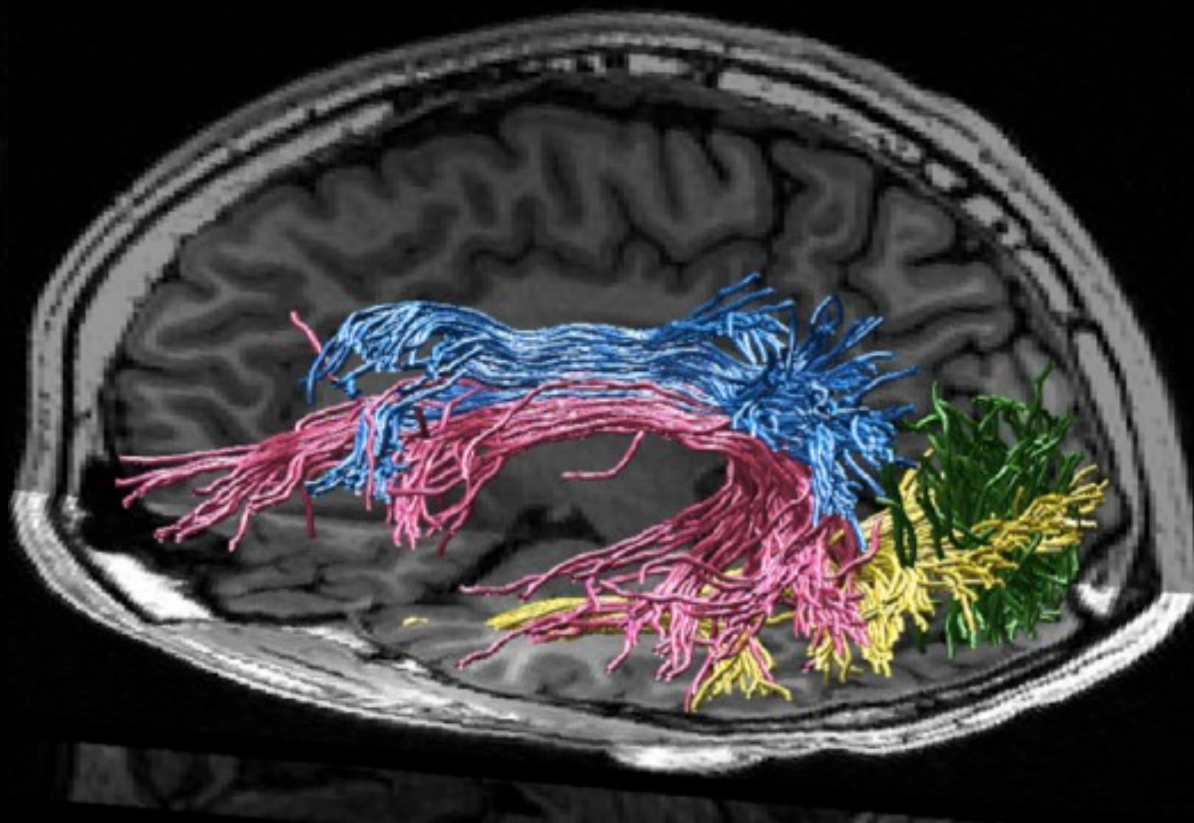


Figure 2. Pathways of Vision and Reading
(A) Major tracts with one termination in the occipital lobe (left)

B



(B) Tissue properties of four white matter tracts are correlated with reading behavior: ILF (yellow), AF (pink), SLF (blue), and VOF (green). These pathways connect cortical regions that are responsive during reading. For clarity, we show only estimated fiber bundles (streamlines) near the core of each tract.