ABSTRACTS

Extraordinary Variations of the Human Mind: Lessons for Anthropogeny

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Our Human Brain: Life on a Continuum

Daniel Geschwind, UCLA

Human behavior and cognition is highly variable and like other human features, consists of many components that are unrelated, each of which is normally distributed in the population. In addition, we know that many of these features are highly heritable, and include factors that may also predispose to common human disorders. The same is true for brain structure, which is the organ that underlies cognition and behavior. Genetic studies show that many of the variants that underlie these phenotypes are common (> 1%) in the population. This supports the view that disease susceptibility is another side of the coin of common human variation, which is largely on a continuum. Some of these disease risks are related to what are considered strengths in other areas, which leads us to a view of human brain function that emphasizes individual differences.

Williams Syndrome: Clues to Links Across Levels from Gene to Cognition

Karen Berman, National Institutes of Health, NIMH

Individuals with Williams syndrome, a fascinating and rare neurogenetic, developmental condition, have only one copy of approximately 25 genes on chromosome 7, instead of the typical, expected two copies (i.e., a "hemideletion"). People with Williams syndrome have a unique profile of striking behavioral features: remarkable hypersociability combined with differential impact on cognitive functions—some mildly affected while others, particularly visuospatial construction, are severely impaired. More recently, individuals with a "duplication" of these same genes, yielding three gene copies, have been studied as well. Interestingly, in contrast people with the Williams syndrome hemideltion, individuals with the Duplication syndrome show a behavioral picture that is in many ways the opposite of that of Williams syndrome itself. In particular, these individuals often have autistic, rather than hypersocial, traits or even autism *per se*, and many have relative strengths in visuospatial processing. Because the genes involved are known, studying persons with Williams syndrome, along with those with the Duplication syndrome, affords a privileged setting for investigating how genes are translated in the brain to produce cognitive and behavioral features.

We have used multimodal brain imaging to identify several fundamental aspects of the brain phenotype in Williams syndrome and the Duplication syndrome that are related to their hallmark behavioral features. We are combining these results with our knowledge of the special genetic characteristic of these populations to test for associations of brain structure and function with "gene-dosage" (one copy of affected genes (i.e., in Williams syndrome) versus two copies (i.e., in the general "control" population) versus three copies. We also test for effects of variation in these chromosome 7 genes in the general population, and we are carrying out longitudinal studies of children with both syndromes in order to understand how brains develop over time in the face of these particular genetic landscapes. More generally, by understanding neurogenetic mechanisms of behavior in the special context of these genetic architectures, information may be obtained about how genetic variation is transduced in the brain to produce the extraordinarily wide range of human variability.

Language at the Extremes

Simon Fisher, Max Planck Institute for Psycholinguistics

The rise of molecular technologies yields exciting new routes for studying the biological foundations of human traits. In particular, researchers have begun to identify genes implicated in unusual disruptions of speech and language skills. My talk will show how genetic studies of language at the extremes can provide powerful entry points into critical neural pathways, using FOXP2 as an example. Rare mutations of this gene cause problems with learning to sequence mouth movements during speech, accompanied by wide-ranging deficits in language production and comprehension. FOXP2 encodes a regulatory protein, a hub in a network of other genes, several

of which have now been associated with language-related impairments. Versions of FOXP2 are found in similar form in many vertebrates; studies of animals and birds suggest it has conserved roles in the development and plasticity of certain sets of neural circuits. Thus, the contributions of this gene to human traits are likely to involve modifications of evolutionarily ancient functions. The FOXP2 story illustrates the value of an interdisciplinary approach for unravelling the complicated connections between genes, neurons, circuits and human cognitive specializations. An exciting prospect for the future is to target speech and language skills at the other extremes of the distribution, identifying genetic factors that contribute to exceptional abilities.

Highly Superior Autobiographical Memory

James McGaugh, UC Irvine

Recent research has identified individuals who have exceptionally strong memories of personal experiences and public events. They are able to recall their activities for most of the days of their lives, after the age of 8 or 10, as well as the days and dates of prominent public events. The presentation will review some of the findings of studies of the memory ability of these individuals and discuss some conceptual and neurobiological implications.

The Incredible Savant Syndrome

Darold Treffert, The Treffert Center

Savant Syndrome is a rare but remarkable condition in which persons with some underlying brain condition, often but not always autism, have some extraordinary islands of ability which stand in stark, jarring contrast to overall limitations. While most cases are congenital and apparent in childhood, recent cases of acquired savant syndrome in which ordinary persons suddenly develop extraordinary musical, artistic, mathematical or memory abilities following head injury, dementia or other CNS incident raise questions about dormant potential within everyone. The challenge is to access that hidden ability without CNS catastrophe.

I met my first savant in 1962. It is impossible to summarize 50+ years of research in 18 minutes. So I have chosen to use clips of Leslie Lemke, the musical savant I know best and have followed for 37 years. His story includes some important aspects of savant syndrome applicable to all savants, congenital or acquired.

All savants “know things they never learned” with access to what is called ‘genetic memory’, the instinctive, inherited knowledge of the rules of music, art of math for example. Leslie Lemke, a prodigious musical savant knows things he never learned (nature) and has never had a music lesson in his life (nuture). He is blind and of course cannot read music. A first film clip demonstrates his instinctive musical playback ability after his initial hearing of Tchaikovsky's first piano concerto. A second clip of Leslie shows him playing a piece he has never heard before *with* someone rather than *after* hearing it. He is parallel processing—receiving, processing, outputting—simultaneously. That is not compatible with a measured IQ of 68 and argues for multiple intelligences. A third clip demonstrates Leslie’s creativity. A final clip addresses the vital role of the family in bringing savant skills to full bloom, and demonstrates the power of music. All savants, followed long enough, proceed on a spectrum of massive recollection to brilliant improvisation to creativity. Savants are not mere tape recorders or copy machines. Savants can be creative.

Until we can explain the savant we cannot fully understand the brain or human potential.

Acquired Savantism in Neurological Conditions

Bruce Miller, UC San Francisco

Visual artistic creativity has been a continual component of human life from the earliest days. Patients with neurodegenerative disease have shown both increased and novel artistic creativity in the face of disease. In Alzheimer’s disease (AD), where the posterior default mode network atrophies, the decline in visuospatial skills tends to lead to more abstract representation and muted color choices. William Utermohlen’s self-portrait series illustrates this progression. On the other hand, frontotemporal dementia (FTD), affects the more frontal salience network and socioemotional systems. The artwork by these patients is often brightly colorful with people or animals in atypical arrangements, reflecting the diminished social or semantic awareness of people and objects in their environment. In patients with left-sided atrophy, decreased language ability may disinhibit the expression of previously dampened visuospatial skills based in the right brain.

Born to be Musical: What We Can Learn from Congenital Anomalies

Isabelle Peretz, Université de Montréal

Congenital anomalies provide a natural experiment—a rare chance to examine the biological basis of musicality by tracing causal links between genes, environment, brain, and behavior. Significant advances have been made on the neurobiology of musicality by studying individuals who were born with severe music-specific disorders. This condition is refered to as congenital amusia (or tone-deafness) and represents the low end of the spectrum.

In contrast, at the other extreme of the spectrum, few scientific studies have sought to delineate the possible underlying neurobiology of musical prodigies. I will present the main and most recent insights that the study of congenital amusia has provided on the biological foundations of musicality and highlight how a similar approach of musical prodigies can reveal the neurobiological foundations of musical giftedness and talent, in general.

Fragments of Genius: Mapping the Mind of a Musical Savant

Adam Ockelford, University of Roehampton

This talk summarizes the findings from a series of experiments undertaken over a number of years with the prodigious musical savant Derek Paravicini. These sought to map his perceptual and cognitive musical abilities with some precision; to identify the strategies he uses in learning new pieces, in re-creating them and in creating new material through improvisation; and to compare these abilities and strategies with those of advanced neurotypical musicians. The research showed that the development of Derek's exceptional musical abilities (in common with all prodigies) were driven by the early acquisition of 'absolute pitch' – the capacity to identify and reproduce notes in isolation from others. Today, this enables him to disaggregate chords with great accuracy and speed, and to learn new pieces quickly and to retain them with a high degree of fidelity over long periods of time. However, Derek's processing of music functions in many respects in the same way as other musicians, with an intuitive understanding of the stylistic grammars through which music is structured. For example, he finds 'tonal' chords easier to disembed than those built up from random combinations of intervals, and, despite his exceptional perceptual abilities, he finds 'atonal' music difficult to memorise and recall, and will 'correct' melodies and chords that did not fit with tonal expectations. In this way, Derek's intuitive, non-conceptual understanding of music appears to function in very much the same way as that of other listeners (with or without musical training). Hence it appears to be the case that, despite his apparently idiosyncratic abilities, Derek (and, in all likelihood other musical savants) in fact function at one extreme of a common continuum of human musicality.

Synesthesia: From Extraordinary Experiences to Enhanced Abilities

Jamie Ward, University of Sussex

People with synaesthesia experience the ordinary world in extraordinary ways: words may have tastes, and music is an audio-visual spectacle. Everybody knows someone with synaesthesia (it affects a few percent of the population) but you may not know who has it because their extraordinary inner world is privately experienced and, to a synaesthete, it is the only reality they know (and so is perfectly ordinary to them!). Having synaesthesia is linked to certain cognitive advantages and this is the focus of this presentation. Synaesthesia is linked to enhanced memory. The enhancements are pervasive (affecting multiple aspects of memory, not just for stimuli that trigger synaesthesia) and meaningful (a 70-year old synaesthete has the memory ability of a 20-year old non-synaesthete). I shall discuss possible mechanisms for how better memory can, in some synaesthetes, lead to truly exceptional memory. In addition to differences in memory, synaesthetes have greater sensory sensitivity on certain objective measures of perception and also in terms of subjective sensitivity (a tendency to find sensory stimuli as aversive). In this regard, some of their symptoms resemble those found in autism spectrum conditions. It has been shown that the autism and synaesthesia co-occur more than expected by chance, and our recent research suggests that this may be particularly apparent in terms of abilities linked to autism (e.g. attention-to-detail) rather than the traditional focus on impairments. Indeed when synaesthesia and autistic tendencies do co-occur they tend to be linked to savant abilities (i.e. exceptional talents).