Autism 101: Clinical and Research Investigations	自閉症第一課: 臨床與研究調查
Clinical and Research Investigations Researchers at UCSF are using many methods to study the autism spectrum disorders. They are isolating genes that may cause crucial differences in social skills, language and behavior. They are using advanced imaging techniques to study the regions of the brain that work differently in individuals with autism and they are using stem cell and other neuroscience techniques to probe explore the autistic brain, one cell at a time. **Stem Cells** Stem cell research is among the cutting-edge techniques that UCSF researchers are using to probe the autistic brain in the search for clues that may help lead to better treatments. At the Institute for Human Genetics, geneticist Lauren Weiss is using patient skin samples to reveal how autism genes affect neural development. These skin cells can be turned into stem cells and then into neurons and other brain cells in order to study their growth, development and function in the lab and develop potential future treatments or preventative strategies. At the Koret Vision Center, neuroscientist Erik Ullian is using stem cells from individuals on the autism spectrum to generate astrocytes, star-shaped cells previously thought to be only gap-fillers in the brain. Ullian's lab is studying the impact of these support cells on how synapses form in the brains of autistic patients, in an effort to uncover the causes of autism on a cellular level.	**臨床與研究調查** 加州大學舊金山分校研究員正利用多種方法以研究在自閉系列領域之內的疾病。他們將逐一分開可能會導致社交技能、語言和行爲重要的差異之基因。他們利用先進的成像技術以研究個別自閉症患者的大腦內工作方式 不同之區域;另外,他們利用幹細胞及其他神經科學技術逐每個細胞去探索自閉症病人之大腦。 **幹細胞** 幹細胞研究是加州大學舊金山分校研究員採用的尖端科技其中之一,用以探索自閉症患者之大腦,尋找線索以協助帶來較佳的治療方法。 在人類遺傳學研究院內,遺傳學家勞倫.魏斯 (Lauren Weiss)正利用患者之皮膚樣本以揭查自閉症基因如何影響神經發育。這些皮膚細胞在實驗室內可以轉化爲幹細胞,然後轉爲神經元及其他腦細胞,可研究其成長、發展及功能,並研發出未來有潛質的療法或防治策略。 在 Koret 視力中心, 神經學家埃里克 (Erik Ullian)正利用取自個別有自閉系列症人士的幹細胞,產生星形膠質細胞,以前這些細胞都被認爲只不過是大腦內用於填充間隙的。Ullian 的實驗室正研究這些支援細胞在自閉症患者的大腦如何形成突觸 (synapses)而產生影響,以盡力在細胞層面上找出自閉症的成因。

Genetics

Researchers at UCSF are using a variety of approaches to investigate the genetic causes of autism.

Dr. Weiss is searching for genetic differences to explain why girls develop autism less often than boys, whether there is interaction between genes and environmental factors in autism, and why only some individuals with certain genetic conditions show autism traits.

Dr. John Rubenstein has investigated the roles of specific genes in regulating how brain cells mature during development. His work aims to explain the mechanisms underlying human neurodevelopmental disorders like epilepsy and autism. Dr. Neil Risch is also collaborating on projects to uncover the genetic causes of autism spectrum disorders.

These genetic approaches may lead to a better understanding of the biology of autism, as well as providing improved tools for risk prediction and diagnosis.

Imaging

Autism spectrum disorders are complex and various. While they are diagnosed by patterns of behavior and language, they usually involve differences in the connections between several regions of the brain. In recent years imaging techniques—such as electroencephalography (EEG) and functional magnetic resonance imaging (fMRI)—have been a useful complement to genetic studies in learning about the causes of autism.

Imaging research indicates that autism involves a

遺傳學

加州大學舊金山分校研究人員正利用多種途徑以探查自閉症之遺傳因素。

魏斯博士正尋找在遺傳上的差異,以解釋不論自閉症有否基因和環境因素之間的相互作用,爲何自閉症在女孩中發生通常較男孩少;而且,爲何只有一些帶有某些基因條件的人才會顯示出自閉症的特徵。

約翰·魯賓斯坦博士 (John Rubenstein)探研於發育過程中一些特定基因如何承擔調節腦細胞成熟之角色。他的目標是要解釋人類神經發育疾病(如癲癇和自閉症)的機制。尼爾·裡舍博士(Neil Risch)也有配合揭開自閉症疾病的遺傳成因之項目。

這些遺傳學之探索方法可能會讓我們更容易理解自閉症的生物學,也可以爲風險預計與診斷提供改良之工具。

成像

自閉系列領域之內的疾病是複雜與多樣的。如果以行爲和語言模式做分析,它們通常包含連接大腦幾個區域之間的的差異。近年,在用基因研究去瞭解自閉症的病因時,成像技術,例如腦電圖(EEG)和功能性磁共振成像(fMRI),已成爲有用的分析附助。

成像研究顯示出自閉症涉及大腦皮層(大腦的外層 --

failure to develop normal connections between regions of the cerebral cortex, an outer layer of the brain where the bodies of the neurons reside. After a period of normal growth, neural circuitry in the cortex begins to lag behind the normal growth pattern, leading to a brain that may lack crucial connections between regions of the cortex. This may account for some of the deficits in language, social skills and behavior that autistic children show. Despite these challenges, people with autism often have more activity in the back of the brain, in regions typically used for visual and spatial perception, and may use these regions to compensate for differences elsewhere in the brain. There are still many questions about the brain differences that lead to the array of symptoms that constitute autism spectrum disorders. Many will be resolved in the coming years, as more imaging studies join with genetics and other research—leading, eventually, to more effective treatments.	神經元停留的地方)區域之間的神經連接未能正常開發。經過一段正常的生長期後,大腦皮層神經迴路的生長開始落後於正常的的生長模式,導致大腦可能缺乏皮質區域之間重要神經元的聯繫。這可以解釋到自閉症兒童在語言、社交技巧與行爲上的一些缺陷。 儘管有這些缺陷的挑戰,但患有自閉症的人在大腦後部和用於視覺和空間知覺的大腦區域經常有較多活動;可以利用這些區域以補償在大腦別處的差異。 現今仍然存在大量關於大腦差異(導致一列的症狀而構成自閉系列領域之內的疾病)的問題。當更多的成像研究參與遺傳學及其他研究,許多問題將會在未來數年內解決到。由此也希望會帶來更爲有效的療法。