Fragile X syndrome is thought to have a significant overlap with autism

Melanie Reid

A genetic defect that causes children to dislike being hugged and sometimes reject all physical affection is closer to being understood following research into the sensory part of the brain.

Scientists at Northwestern University, Illinois, and the University of Edinburgh explored fragile X syndrome — a genetic condition associated with an extreme hypersensitivity to sounds, touch, smells and visual stimuli that can result in social withdrawal, hyperarousal or anxiety. Some sufferers are even hypersensitive to material on their skin.

The scientists found that critical phases in the brain's development may be mistimed in people with the condition. This may result in inappropriate communication between brain cells.

By recording electrical signals in the brains of mice, bred to mimic the condition, the researchers found that connections in the brain's sensory cortex were late to mature. The study, published in the journal *Neuron*, found that these changes in the brain's connections occurred much earlier than previously thought — in the first week post-natal in mice, which is equivalent to the middle of the second trimester of pregnancy.

The mistiming, in which the neurons do not communicate properly, may cause further problems with the correct wiring of the brain. The hope is that by understanding how and when the function of the brain is affected in fragile X, a therapy may become possible.

"There is a critical period during development when the brain is very plastic and is changing rapidly," said Anis Contractor, from the Feinberg School of Medicine at Northwestern University. "All the elements of this rapid development have to be co-ordinated so that the brain becomes wired correctly and therefore functions properly."

People with the syndrome have debilitating sensory as well as cognitive problems. "They have tactile defensiveness," Dr Contractor said. "They don't look in people's eyes, they won't hug their parents and they are hypersensitive to touch and sound. All of this causes anxiety for family and friends as well as for the fragile X patients themselves."

Peter Kind, who led the study at the University of Edinburgh, said: "We know there are key windows during which the brain develops, both in the womb and afterwards. The general principle is that if these time windows have shifted, then that could explain the cognitive problems."

Professor Kind said that this could be demonstrated by the fact that a child with a cataract which was not corrected
would become permanently blind in the eye, whereas an adult would be able to regain their sight after an operation.

“We’ve learnt that these changes happen much earlier than previously thought, which gives valuable insight into when we should begin therapeutic intervention for people with these conditions,” he said. “It also has implications for the treatment of autism since the changes in the brains of fragile X and autistic people are thought to significantly overlap.”

Fragile X syndrome is as common as cystic fibrosis, affecting about 1 in 4,000 males and 1 in 8,000 females worldwide. The Fragile X Society believes that there are many people who have not been diagnosed. It shows up in early infancy and progressively worsens throughout childhood, causing intellectual disability as well as social, language and behavioural problems.

Fragile X syndrome is caused by a gene mutation in the X chromosome that interferes in the production of a protein called fragile X mental retardation protein. Fragile X is so named because the X chromosome appears broken or kinked.

Tim Potter, from the Fragile X Society, said: “We welcome any research that helps us understand fragile X and which may open the way to reversing the effects or preventing them ever happening.”

Contact our advertising team for advertising and sponsorship in Times Online, The Times and The Sunday Times, or place your advertisement.

Times Online Services: Dating | Jobs | Property Search | Used Cars | Holidays | Births, Marriages, Deaths | Subscriptions | E-paper

News International associated websites: Milkround

Copyright 2010 Times Newspapers Ltd.
This service is provided on Times Newspapers' standard Terms and Conditions. Please read our Privacy Policy. To inquire about a licence to reproduce material from Times Online, The Times or The Sunday Times, click here. This website is published by a member of the News International Group. News International Limited, 1 Virginia St, London E98 1XY, is the holding company for the News International group and is registered in England No 81701. VAT number GB 243 8054 69.