THE ROYAL SOCIETY

Translating the biology of intellectual disability and autism

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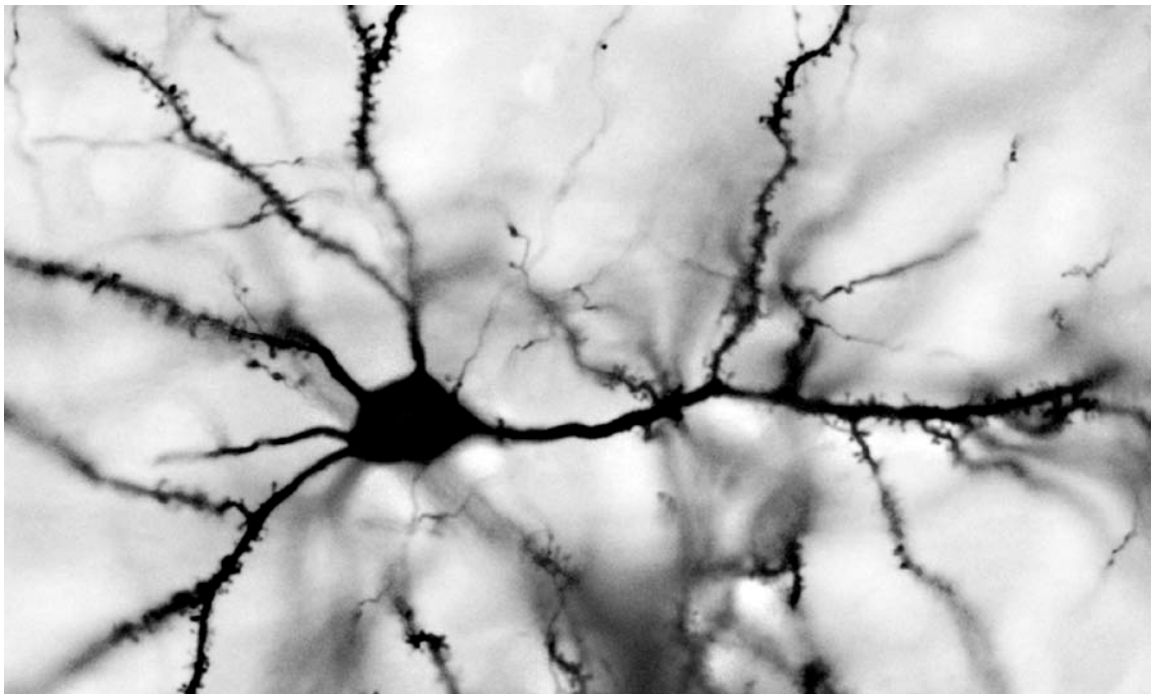
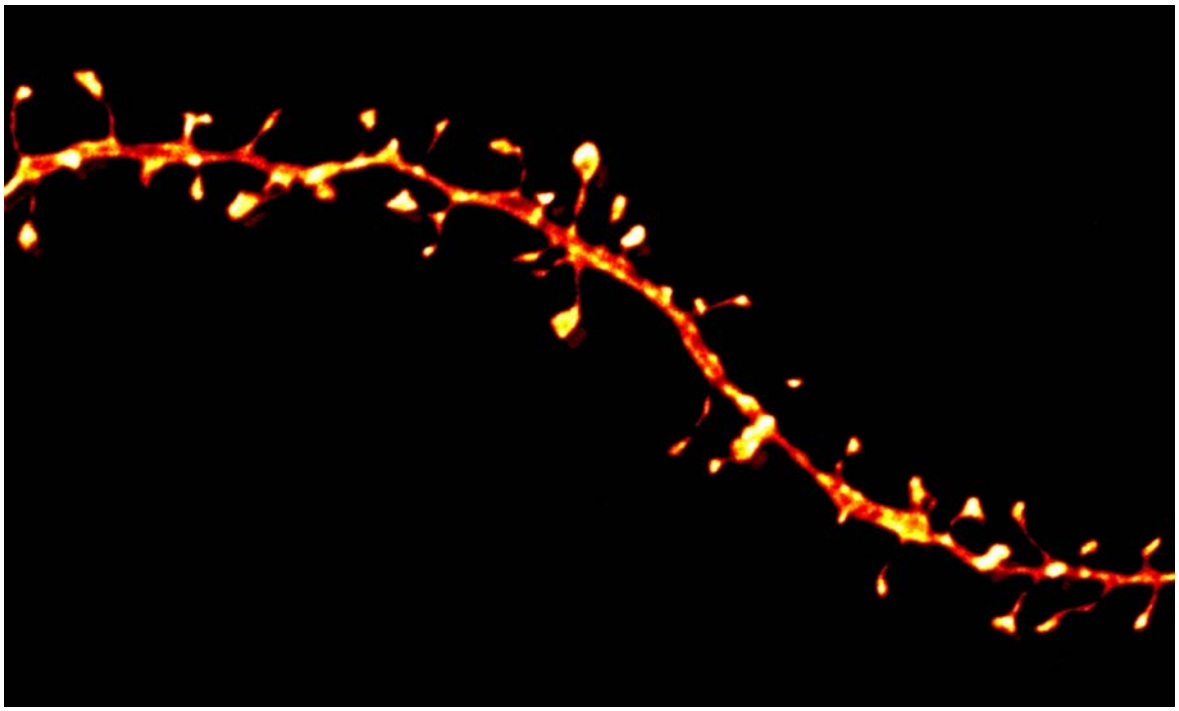
Based at Edinburgh University, the Patrick Wild Centre is dedicated to finding out the biological mechanisms behind and testing new treatments for many forms of intellectual disability and autism. Charged with the aim of bridging the gap between laboratory neuroscience and clinical innovation, the Centre was initially set-up through the combination of a fundraising initiative led by the parents of an affected child and a generous donation from Dr Alfred Wild, whose brother Patrick was severely disabled by autism.

In order for the brain to function effectively, the billions of nerve cells which it contains must communicate efficiently with each other. This communication occurs across a gap between nerve cells called the synapse. Any disruption to the function of a synapse can cause problems with this communication and potentially lead to some forms of intellectual disability and autism. Therefore, understanding the complex science behind synaptic function is a key step to developing new therapies for people with these conditions.

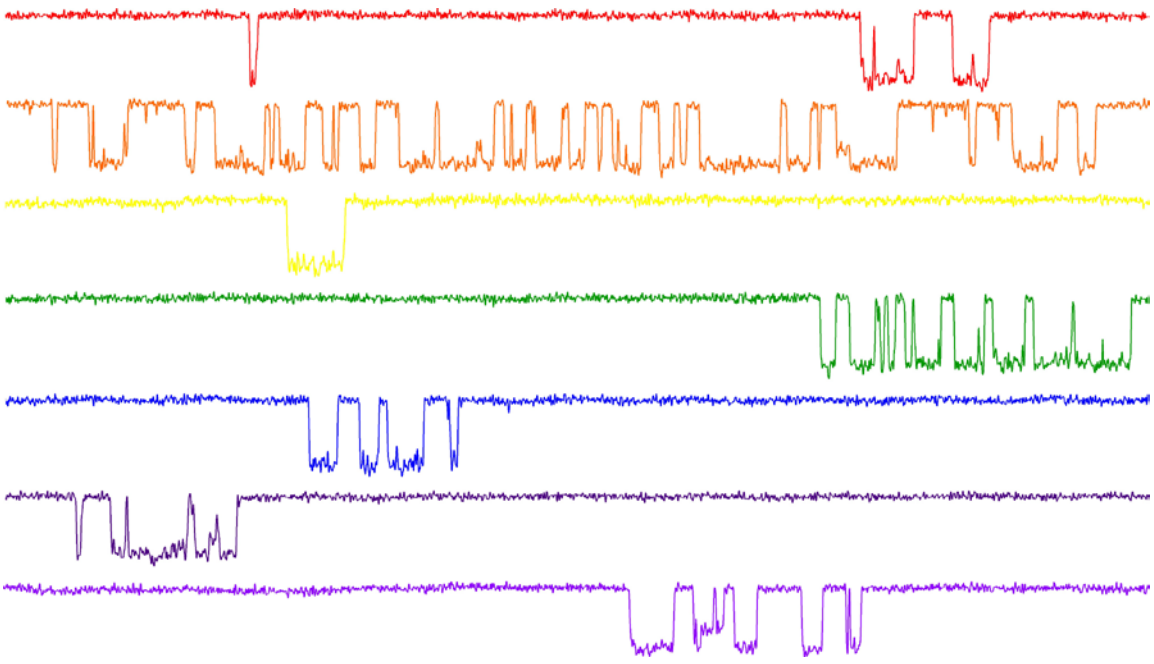


Photograph 1: A typical nerve cell from the brain showing the branches (dendrites) upon which other nerve cells connect.



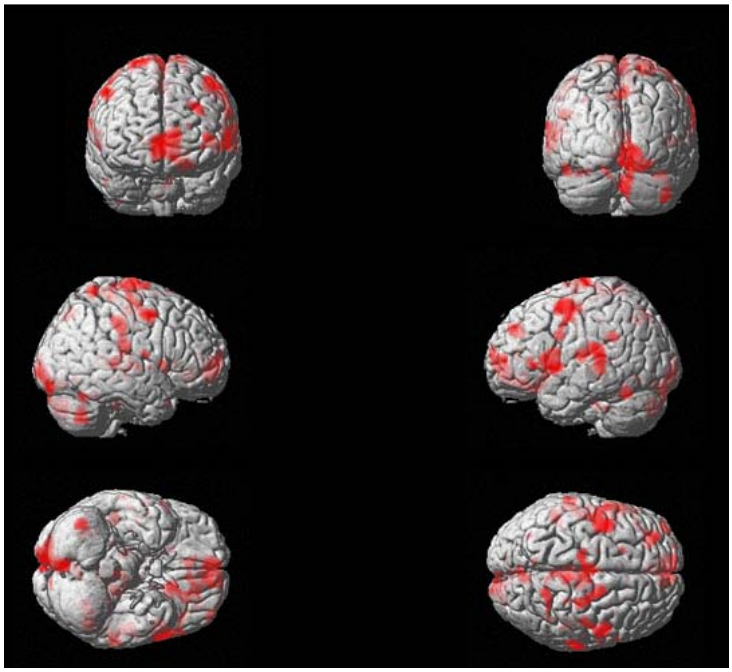
Photograph 2: A close up of a dendrite. The mushroom shaped spines are where this dendrite connects to another nerve cell

However, understanding how the synapse functions in the laboratory is not enough. The next step is to take findings from the laboratory and look for parallel findings in real people affected by these conditions – a process often referred to as 'translation'. Although it is not generally possible to observe or examine the function of nerve cells in the brain of an awake and conscious person in the level of detail that one can in a laboratory, the Patrick Wild Centre is using advances in brain imaging techniques to draw comparisons between their findings in people and those from the laboratory.



Photograph 3: Electrical recordings taken from a nerve cell in the laboratory

One of the main conditions which the Centre is working on is fragile X syndrome, a single gene disorder which is commonly associated with both autism and intellectual disability. New treatments for this condition have been developed through an understanding of dysfunction of the fragile X gene affects the synapse and the Patrick Wild Centre is part of several multinational efforts to test these in affected people. It is also hoped that, by understanding how therapies affect brain function in the laboratory and in people, it will be possible to use them to develop more treatments for both fragile X syndrome and other forms of intellectual disability and autism.



Photograph 4: Brain activity in an individual with fragile X syndrome

Each aspect of this research is highly technical and requires the collaboration of many scientists working across the University of Edinburgh and beyond. Bringing this kind of collaborative scientific effort to bear on complex problems of disability is one way of improving the lives of people affected by these conditions and their families.