IMP Press Release

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13. December 2012

Embargoed until Dec. 13, 2012, 18.00 MEZ

A Key Gene for Brain Development

Neurobiologists at the Research institute of Molecular Pathology (IMP) in Vienna have discovered one of the key genes required to make a brain. Mutations in this gene, called TUBB5, cause neurodevelopmental disease in children.

About one in ten thousand babies is born with an abnormally small head. The cause for this disorder – which is known as microcephaly – is a defect in the develoment of the embryonic brain. Children with microcephaly are severely retarded and their life expectancy is low. Certain cases of autism and schizophrenia are also associated with the dysregulation of brain size.

The causes underlying impaired brain development can be environmental stress (such as alcohol abuse or radiation) or viral infections (such as rubella) during pregnancy. In many cases, however, a mutant gene causes the problem.

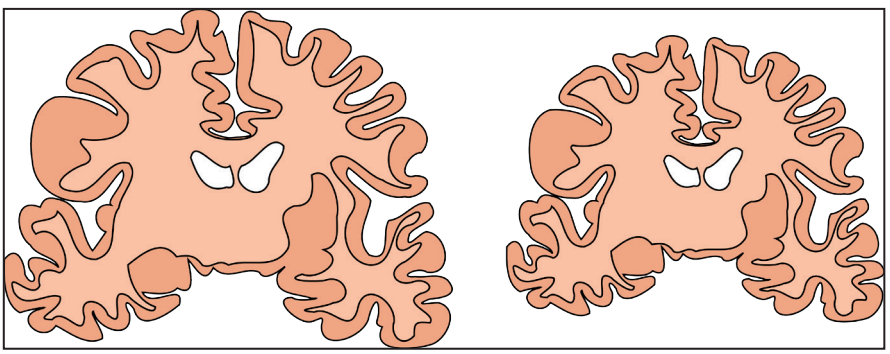
David Keays, a group leader at the IMP, has now found a new gene which is responsible for microcephaly. Together with his PhD-student Martin Breuss, he was able to identify TUBB5 as the culprit. The gene is responsible for making tubulins, the building blocks of the cell's internal skeleton. Whenever a cell moves or divides, it relies on guidance from this internal structure, acting like a scaffold.

The IMP-researchers, together with collaborators at Monash University (Victoria, Australia), were able to interfere with the function of the TUBB5 in the brains of unborn mice. This led to massive disturbances in the stem cell population and impaired the migration of nerve cells. Both, the generation of large numbers of neurons from the stem cell reservoir and their correct positioning in the cortex, are essential for the development of the mammalian brain.

To determine whether the findings are also relevant in humans, David Keays collaborates with clinicians from the Paris-Sorbonne University. The French team led by Jamel Chelly, examined 120 patients with pathological brain structures and severe disabilities. Three of the children were found to have a mutated TUBB5-gene.

This information will prove vital to doctors treating children with brain disease. It will allow the development of new genetic tests which will form the basis of genetic counseling, helping parents plan for the future. By understanding how different genes cause brain disorders, it is hoped that one day scientists will be able to create new drugs and therapies to treat them.

The new findings by the IMP-researchers are published in the current issue of the journal "Cell Reports". For David Keays, understanding the function of TUBB5 is the key to understanding brain development. "Our project shows how research in the lab can help improve lives in the clinic", he adds.



Comparison of the size of a normal brain (left) and a microcephalic brain (right). Drawing based on coronal sections of human brains (Copyright: IMP)

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The paper „Mutations in the β-tubulin Gene TUBB5 Cause Microcephaly with Structural Brain Abnormalities“ is published on December 13, 2012, in the online-Journal Cell Reports.

Illustrations to be used free of charge can be downloaded from the IMP-Website: *http://www.imp.ac.at/pressefoto-microcephaly*

About the IMP

The Research Institute of Molecular Pathology (IMP) in Vienna is a basic biomedical research institute largely sponsored by Boehringer Ingelheim. With over 200 scientists from 30 nations, the IMP is committed to scientific discovery of fundamental molecular and cellular mechanisms underlying complex biological phenomena. Research areas include cell and molecular biology, neurobiology, disease mechanisms and computational biology. The IMP is a founding member of the Campus Vienna Biocenter.

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