ÖAW AUSTRIAN ACADEMY OF SCIENCES

IMBA - THE VIENNA DBA PROJECT

WHAT IS DBA

Diamond Blackfan anaemia is a hereditary form of a serious chronic blood disorder caused by a congenital defect that has yet to be pinpointed. It impairs the formation of red blood cells in the bone marrow. DBA is a very rare disease that affects 5 - 7 out of every million live births. In most cases the disease manifests sporadically, but some patients pass it on, meaning that other family members may be affected. DBA patients have no or only few maturing progenitor cells for red blood cells.

JOSEFINE AND LUIS: TWO VERY SPECIAL CHILDREN

In Vienna, ten children are affected. Two of them, Josefine and Luis, are brother and sister. Their case is particularly interesting from a scientific perspective, since one of their parents carries the same mutation but is unaffected by the condition. Researchers at IMBA (Institute of Molecular Biotechnology of the Austrian Academy of Sciences), led by Prof. Josef Penninger, are using cutting research tools to study why individuals carrying certain RPS gene mutations can develop the DBA disease, whereas another one with the same mutation may not develop the disorder. Precisely, to elucidate the genetic cause of DBA disease and genes which are particularly resistant to the disease known as "supergenes". The latter prevent people from falling ill despite their genetic disposition. There is a possibility that the same mechanisms may play a role in DBA, and also in cancer.

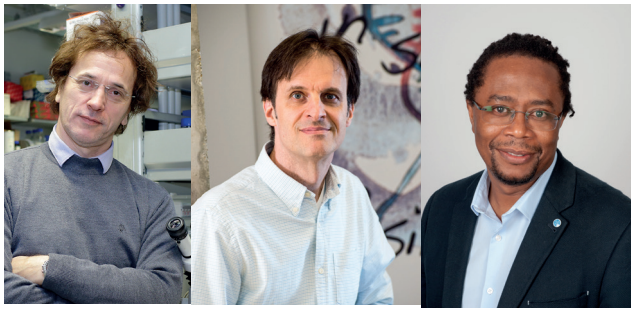
RESEARCH BENEFITS EVERYONE

The knowledge gained from this study will benefit the general population, facilitating the translation of rare disease research into tangible treatments applicable in various areas of personalized medicine be hopefully useful to children like Josefine and Luis.

ABOUT IMBA

IMBA (Institute for Molecular Biotechnology) is one of Europe's leading centres for basic biomedical research and the largest institute of the Austrian Academy of Sciences. Among other awards, researchers at IMBA have won 12 ERC Grants from the European Research Council and three Wittgenstein Prizes (the highest Austrian science prize).
Its research focuses include: cancer, heart regeneration, stem cells, brain disease, osteoporosis, egg cells and female fertility, infectious diseases and rare diseases.

www.imba.oeaw.ac.at



The scientific leading team of the Vienna DBA project:
Prof. Josef Penninger, IMBA
Prof. Javier Martinez, Medical University Vienna
Dr. Chukwuma Allison Agu, IMBA Stem Cell Core Facility

IMBA Institute of Molecular Biotechnology of the Austrian Academy of Sciences

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PARENTS EXPLAIN

"Josefine (5) and Luis (6) have both suffered from Diamond Blackfan anaemia from birth. This means that they are dependent on transfusions and have to take some pretty heavy medication to keep the iron overload in check. Josefine and Luis are unable to produce red blood cells, which means that they have to go to the "blood filling station" at St. Anna Children's Hospital Vienna for a transfusion every three weeks.
It's clear that this disease is caused by a genetic mutation that is associated with certain phenotypic characteristics. But it is still not clear exactly why this disease is triggered. Owing to its rarity – there are only 5-7 cases per million – work has already started on setting up internationally uniform diagnostic standards and registration procedures to collate data on the disease's progression.
Research into what actually triggers DBA is still in its infancy. We want to do everything that we can to help find out what causes DBA, not just out of our love for our children, but because we firmly believe that research into its origins could deliver significant insights into other types of bone marrow disease.
We know from the literature that in a great many cases research into diseases start with the individuals affected and their dogged determination not to simply accept the status quo. We want to take inspiration from all of these cases and firmly believe that by working with Josef Penninger and his team, we will be able to achieve something truly groundbreaking for science and in the field of research into rare diseases.
And of course we want to find a solution that stops our children Josefine and Luis' suffering." (Josefine and Luis' parents)

THE VIENNA DBA PROJECT AT IMBA

In cooperation, the working groups led by Prof. Josef Penninger (IMBA), Dr. Chukwuma Agu (IMBA Stem Cell Core Facility), and Prof. Javier Martinez (Medical University of Vienna) are attempting to identify the molecular mechanisms behind DBA which lead to the onset of the illness. Blood samples are being taken from the affected families for derivation of mononuclear blood cells. Afterwards, these cells are used for the generation of induced pluripotent stem (iPS) cells. Discreet erythroid progenitors will be generated from the iPS cells and characterized. We hope that this project will provide important insights into the DBA disease mechanism and the maturation process involved in the formation of red blood cells that is inhibited in patients. Molecular-level understanding is a key part of the development of a possible therapy. The scientists regularly exchanges their experiences with Dr. Leo Kager, Associate Professor of Pediatrics at St. Anna Children's Hospital in Vienna, Medical University of Vienna.

The initiators of the Vienna DBA Project: Prof. Josef Penninger (left) with Marianne and Boris Marte, parents of two children with DBA

HOW YOU CAN HELP

The DBA project is planned for three years (2017-2019) and requires € 210,000.- per year.
So far, more than € 150.000.- have been raised by IMBA Fundraising and the Marte family. For more information please contact Sylvia Weinzettl at IMBA: *sylvia.weinzettl@imba.oeaw.ac.at*

Please join the more than 230 donors so far and support our work!

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Every contribution is welcome. Thank you for your support!

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