Initial Lay Summary

Androglobin and primary ciliary dyskinesia

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Originaltitel: Linking the newly identified androglobin to pulmonary and tracheal ciliary function

Main applicant: Dr. Keppner Anna, University of Fribourg Research management

Synopsis

Androglobin (Adgb) is the most recent member of the globin family, which includes also hemoglobin and myoglobin. Genetic deletion of Adgb in mice leads to a disease called primary ciliary dyskinesia (PCD). This study aims at identifying the mechanistic link between Adgb and the pulmonary defects observed in PCD, such as mucus accumulations and chronic bronchitis.

Dr. Anna Keppner, principal investigator of the study

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Context

Primary ciliary dyskinesia (PCD) is a genetic disease affecting cilia, which are small hair-like antennas found on almost every cell type, and whose main function is to drain fluids, such as mucus in the lungs or cerebrospinal fluid in the brain. Patients suffering from PCD display symptoms such as chronic bronchitis, mucus accumulations in the lungs and sinuses, but may also display infertility, brain swelling, and cardiac defects. Our research focuses on androglobin, the most recently identified member of the globin family, which also includes hemoglobin and myoglobin. Mice genetically lacking Adgb develop PCD, suggesting a function of Adgb in cilia function.

Objectives and methods

In this study, we aim at identifying how Adgb is linked to cilia, and how its genetic absence leads to PCD. To achieve this goal, we will isolate and culture cells from the trachea of control and Adgb-deficient mice, in order to visualize the cilia, to measure their motility, and to identify the underlying biological defect leading to the disease. The cells will then be treated using different pharmacological compounds, in order to get a better understanding on how Adgb is influencing the cilia. PCD patients suffer from breathing difficulties, mostly due to the mucus accumulations in the lungs. Therefore, this project also aims at measuring the breathing capacity of control and Adgb-deficient mice, using a technique called whole-body plethysmography.

Significance

The diagnosis of PCD mostly relies on genetic testing, but current tests can only identify defects in less than 70% of definite PCD cases. Thus, identifying Adgb as causative for the disease will enhance the understanding of the disease, and will contribute to potential novel therapeutic approaches to treat PCD.

Start and duration

The project will start in September 2022, and should last one year.

	Betrag	
Total research budget	CHF	96'000
Grants promised / received by third parties	CHF	0
Grants pending from third parties	CHF	0
Grants being sought from the Swiss Lung Association	CHF	96'000
Amount to be acquired by researchers	CHF	16'000
Contribution from Research Fund of the Lung Association	CHF	24'000
Donations required from third parties	**CHF**	**56'000**