Myotonic Dystrophy FOUNDATION

English Translation of Dr. Bruno Fleischer's "On Myotonic Dystrophy with Cataracts: A Hereditary, Familial Degeneration"

The Myotonic Dystrophy Foundation (MDF) facilitated the translation of Dr. Bruno Fleischer's systematic genealogic analysis of patients and multigenerational families with myotonic dystrophy (DM). This work, entitled "About Myotonic Dystrophy with Cataracts", was originally published 1918 in Fleischer's native German language.

"Über myotonische Dystrophie mit Katarakt: Eine hereditäre, familiäre Degeneration"; May 1918, Albrecht von Graefes Archiv für Ophthalmologie, Volume 96, pages 91–133, (1918).

Beginning 1901, Dr. Schleicher, then a professor and attending physician at the University Ophthalmology Clinic at the University of Augsburg, Germany, systematically performed family history and genealogic investigations of multigenerational family pedigrees of patients with myotonic dystrophy and/or cataracts and reexamined cases seen in his hospital to establish the presentation of cataracts prior to the muscular and neuromuscular presentation of DM.

The work is of seminal importance for myotonic dystrophy as:

- It clearly establishes the disease as hereditary and also as progressive,
- Non-age related cataracts are frequently unambiguous and the only manifestation of the disease, and
- Cataracts precede, generationally, the presentation of neuromuscular phenotypes.

Dr. Fleischer's work is often cited as a foundational for the concept of *genetic anticipation*, a genetic phenomenon where symptoms of a disease become more severe and appear at an earlier age in each generation, most commonly due to the hereditary nature of the disease.

The study is also of interest to the geneticist and student in genetics, showcasing the importance of family history, the importance of thorough pedigree analysis, and the subsequent combination of family history analysis with the integration of anecdotal evidence.

Fleischer's original article was translated by Dr. Corinna Fehle, PhD, with funding from the Myotonic Dystrophy Foundation. It was thereafter clinically and scientifically reviewed by Dr. Benedikt Schoser, MD and Dr. Andy Rohrwasser, PhD. We would like to thank Dr. Darren Monckton for the idea to complete this translation.

The translation is available upon request (info@myotonic.org). We emphasize to the interested reader the importance of the actual family pedigrees of the multigeneration families present in the PDF of the original article.