Cause of hereditary blindness discovered

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Initially the occurrence of progressive retinal degeneration - progressive retinal atrophy, in man called retinitis pigmentosa - had been identified in Schapendoes dogs. Retinitis pigmentosa is the most common hereditary disease which causes blindness in humans. The researchers report on their findings, in *Human Molecular Genetics*.

Based on the new findings, the researchers from Bochum have developed a genetic test for diagnosis in this breed of dogs that can also be used predictively in breeding. Schapendoes dogs are originally a Dutch breed of herding dog, which is now kept mainly in Holland, Germany, Northern Europe and North America. However, the research results are also potentially significant for people. The scientists are currently investigating whether mutations of the CCDC66 gene could also be responsible for some retinitis pigmentosa patients.

"Since at the beginning of the work, the importance of the CCDC66 protein in the organism was completely unknown, in collaboration with Dr. Thomas Rülicke (Vienna) and Prof. Dr. Saleh Ibrahim (Lübeck), we developed a mouse model with a defect in the corresponding gene" explained Prof. Epplen. The aim was initially to obtain basic information about the consequences of the CCDC66 deficiency in order to draw conclusions on the physiological function of the protein. "Fortunately, the mice showed exactly the expected defect of slow progressive impaired vision", said Epplen. "Along with Dr. Elisabeth Petrasch-Parwez (RUB) and Prof. Dr. Jan Kremers (Erlangen), we were able to anatomically and functionally study the entire development of the visual defect in the mouse in just a few months, whereas the progress takes

years in humans and dogs." In this interdisciplinary project, the researchers have precisely documented and characterised the progress of retinal degeneration. Epplen: "Interestingly, the CCDC66 protein is, for example, only localised in certain structures of the rods".

The insights gained from the studies of the working group can now be applied in order to better understand the processes that cause this inherited disorder. The mouse model will be studied further, as the researchers said: "with regard to malfunctions of the brain, but naturally, above all as a prerequisite for future therapeutic trials in retinitis pigmentosa."

More information: Ccdc66 null mutation causes retinal degeneration and dysfunction. Gerding WM, Schreiber S, Schulte-Middelmann T, de Castro Marques A, Atorf J, Akkad DA, Dekomien G, Kremers J, Dermietzel R, Gal A, Rülicke T, Ibrahim S, Epplen JT, Petrasch-Parwez E. Hum. Mol. Genet. (2011) first published online June 16, 2011 doi:10.1093/hmg/ddr282

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