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Pathogenesis of mutant myocilin-related primary open angle glaucoma

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Glaucoma affected about 66.8 million people worldwide. Primary open angle glaucoma (POAG) contributed to 70% cases of glaucoma. Of different genetic type discoveries in glaucoma, MYOC gene mutation has been considered as a single POAG-causing gene. This mutation caused production of mutant myocilin protein which was rarely observed in people without glaucoma. An exact pathogenesis of how mutant myocilin induced POAG is important for further intervention and treatment, meanwhile it remains unknown. This literature review is aimed to describe the pathogenesis of POAG related to mutant myocilin. Reviewed literatures revealed that turnover of mutant myocilin involved autophagy pathway, rather than ubiquitin-proteasome and lysosomal pathways which was involved in turnover of non-mutant Myocilin. Grp94 was found as a product of unfolded protein response to mutant myocilin. Grp94 bound mutant myocilin and directed its degradation to endoplasmic reticulum-associated degradation (ERAD). ERAD pathway degraded mutant myocilin inefficiently, which resulted in accumulation of mutant myocilin. The accumulation of mutant myocilin in trabecular meshwork (TM) cells led to ER stress-induced cell death. TM cell death interfered the outflow of aqueous humour therefore increased intra-ocular pressure. Another research revealed that mutant myocilin is consistently causing stress-induced cell death by increasing sensitivity towards oxidative stress. Knowledge of the mutant myocilin involvement in POAG can help further investigation strategies for myocilin-related glaucoma. In summary, the binding between Grp94 and mutant myocilin induced the death of TM cell and led to the development of POAG.

Biography

Putri Pamulani is a Medical Student from Universitas Padjadjaran. Talk about ideas, interested in medical science and interpersonal skill, as well as social and cultural activity, guitar and amateur art and ceramic, opened to new things and dream to explore the earth!

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