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Abstract: Long-term follow up of retinal angiomatosis – a retrospecitve ophthalmological study

Purpose: To evaluate the clinical course of retinal angiomatosis and to determine the correlation between retinal angioma and von Hippel Lindau disease (VHL): Furthermore to give recommendations for an ophthalmological screening.
Another aim was to distinguish between sporadic and hereditary angiomas and to examine the relationship between retinal manifestations and other typical organic manifestations of VHL.

Methods: 63 patients with retinal angiomatosis were examined in terms of the number and size of angiomas and visual function. In addition a work up for other VHL lesions and molecular genetic testing was performed. The diagnosis of VHL was based on personal and family history, the presence of other VHL associated organ lesions or the presence of a mutation of the VHL gene.

Results: Mean follow up was 5,8 years. The calculated prevalence for bilateral retinal angiomatosis was 100% at age 56.4 years. The prevalence of legeal blindness due to retinal angiomas was calculated at 41% of all eyes by age 61,1 years. Legal blindness occured at a mean age of 23,2 years: risk faktors included large angiomas, manifestation at an early age or symptomatic angiomatosis. Analysis of growth behaviour showed that large angiomas which become symptomatic in young adults, start growing in childhood. In the case of uncomplicated angiomatosis the development of new angiomas was generally slow and only small angiomas were detected during regular follow-up examinations. Larger angiomas were however observed in those eyes showing multiple retinal angiomas or retinal detachment.
In our group of patients we found all of the typical clinical manifestations of VHL. The number of retinal angiomas per patient showed a positive correlation to the number of other organs affected by VHL disease.

Multiple retinal angiomas always featured with VHL. Thus single angiomas can be sporadic or the first retinal manifestation of VHL and further examination ought to follow. Moleculagenetic testing was particulary effective here.

Conclusion: Retinal angiomatosis in VHL disease bears a high risk of severe vision loss at an early age. Lifelong ocular screening for presymptomatic lesions starting at pre-school age is therefore essential after gene-carrier status has been determined.