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Risk factors for survival in patients with von HippelLindau disease

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Introduction & Objectives

Historically, von Hippel-Lindau (VHL) disease is characterized by a poor survival. Although genotype–phenotype correlation has been described in many studies, the risk factors for VHL survival remain unclear. This study aims to evaluate the median survival of Chinese patients with VHL disease and explore whether VHL survival is influenced by genetic and clinical factors.

Materials & Methods

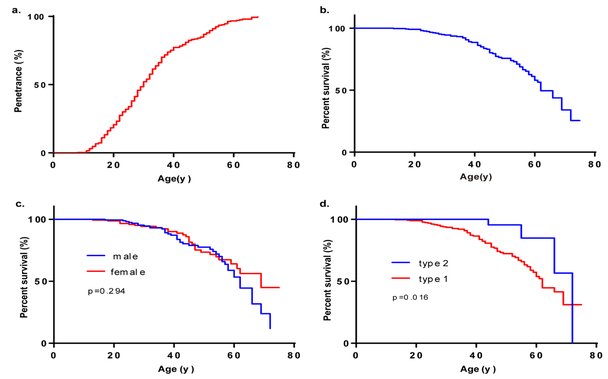
In this retrospective study, we recruited 340 patients from 127 VHL families. Kaplan-Meier plot and Cox regression model were used to evaluate the median survival and assess how survival was influenced by birth year, birth order, sex, family history, mutation type, onset age and first presenting symptom.

Results & Conclusions

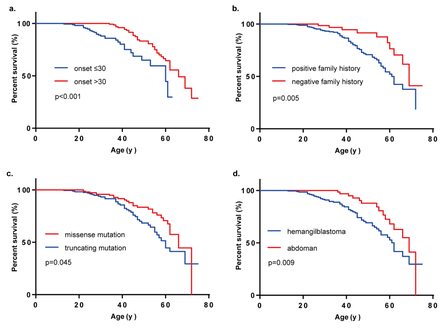
1. Characteristics of the patients included in the survival analyses.

	No. of patients		No. of patients
Overall	340	**Onset age**	
Sex		≤30y	162(54.4%)
male	179(52.6%)	>30y	136(45.6%)
female	161(47.4%)	**First lesion**	
Birth year		CHB	157(52.7%)
≤1950	36(10.6%)	RA	37(12.4%)
>1950	304(89.4%)	RCC	52(17.4%)
Birth order		PHEO	19(6.4%)
1	151(44.4%)	PCT	16(5.4%)
≥2	189(55.6%)	Unknown	17(5.7%)
Family history		**Death and cause**	
yes	270(79.4%)	death(any cause)	72
no	70(20.6%)	CHB	48(67.7%)
Mutation type		RCC	20(27.8%)
missense	165(48.5%)	PHEO	2(2.8%)
truncating	175(51.5%)	Non-VHL related	2(2.8%)
Onset			
affected	298(87.6%)		
not affected	42(12.4%)		

2. Survival of Chinese patients with VHL disease.



3. Patients with early onset age, positive family history and truncating mutation types had poorer overall survival.



4. Risk factors for survival in patients with VHL disease.

Variables	Univariate analyses			Multivariate analyses		
	HR	95% CI	P value	HR	95% CI	P value
Onset age (≤30y vs. >30y)	3.202	1.904-5.385	**<0.001**	2.699	1.461-4.988	**0.002**
Sex (male vs. female)	1.318	0.815-2.132	0.261	1.321	0.798-2.186	0.279
Birth year	1.011	0.988-1.036	0.346	0.992	0.967-1.018	0.565
Birth order (first vs. others)	0.943	0.562-1.582	0.824	0.829	0.490-1.404	0.486
Family history (yes vs. no)	2.143	1.110-4.137	**0.023**	2.519	1.275-4.980	**0.008**
Mutation (missense vs. truncating)	0.600	0.366-0.983	**0.042**	0.589	0.354-0.980	**0.042**
First symptom (HB vs. abdomen)	2.109	1.188-3.747	**0.011**	1.535	0.792-2.973	0.204

Abbreviations: HB, hemangioblastoma

5. Conclusions: This largest VHL survival analysis indicates that onset age, family history, mutation type and first presenting symptom have an effect on the survival of patients with VHL disease, which is helpful to genetic counselling and clinical decision-making.