



## 2013

166 2013 2<sup>7)</sup> 2001 1 2011 3 SEGA<sup>1</sup> 2<sup>2</sup> 63%<sup>1</sup> 20<sup>1</sup> 42<sup>3</sup> 65<sup>1</sup> TSC2<sup>1</sup> 32<sup>8)-11)</sup> 1 p<0.0001 2 p=0.0005  $\chi^2$  93<sup>3</sup> 83%<sup>3</sup> NMI<sup>3</sup> 39% 3 p<0.0001  $\chi^2$

### 2

	95% CI	p	
SEGA	77	70-84	50-100 1.00 ND
SEGA	2	0-5	10-35 <0.0001 L
	63	56-70	75-95 0.0005 L
	20	14-26	50 <0.0001 L
	42	34-49	65-75 <0.0001 L
Level 3	3.6	1-6	
Level 2	21	15-27	
Level 1	17	11-23	
	21	15-27	25-50 0.24 ND
	71	64-78	64-78
	61	53-69	45-55 1.00 ND
>4cm	29	22-36	
	28	21-35	15-32 0.65 ND
	2.6	0-5	2 0.59 ND
	79	71-87	
LAM	39	29-49	2-40 0.92 ND
MMPH	71	62-80	
	98.8	97-100	
	93	89-97	75-80 <0.0001 H
	46	38-54	12-40 0.13 ND
(>3)	65	58-72	>90 <0.0001 L
	83	77-89	20-57 <0.0001 H
	64	57-71	15-80 1.00 ND
	49	39-59	

Pathology	%	95% CI	n	p	Significance
Overall	46	37-55	40-60	0.90	ND
<b>Pathology &gt;20</b>					
>20	57	43-71			
≤20	28	14-42			
<b>Pathology</b>					
≤20	27	18-36			
<b>Pathology</b>					
TSC1	28	18-38	20	0.08	ND
TSC2	32	21-43	66	<0.0001	L
NMI	39	30-52	20	<0.0001	H

SEN  
 SEGA  
 LAM  
 MMPH multifocal micronodular pneumocyte hyperplasia  
 CI  
 H  
 L  
 ND  
 $\chi^2$

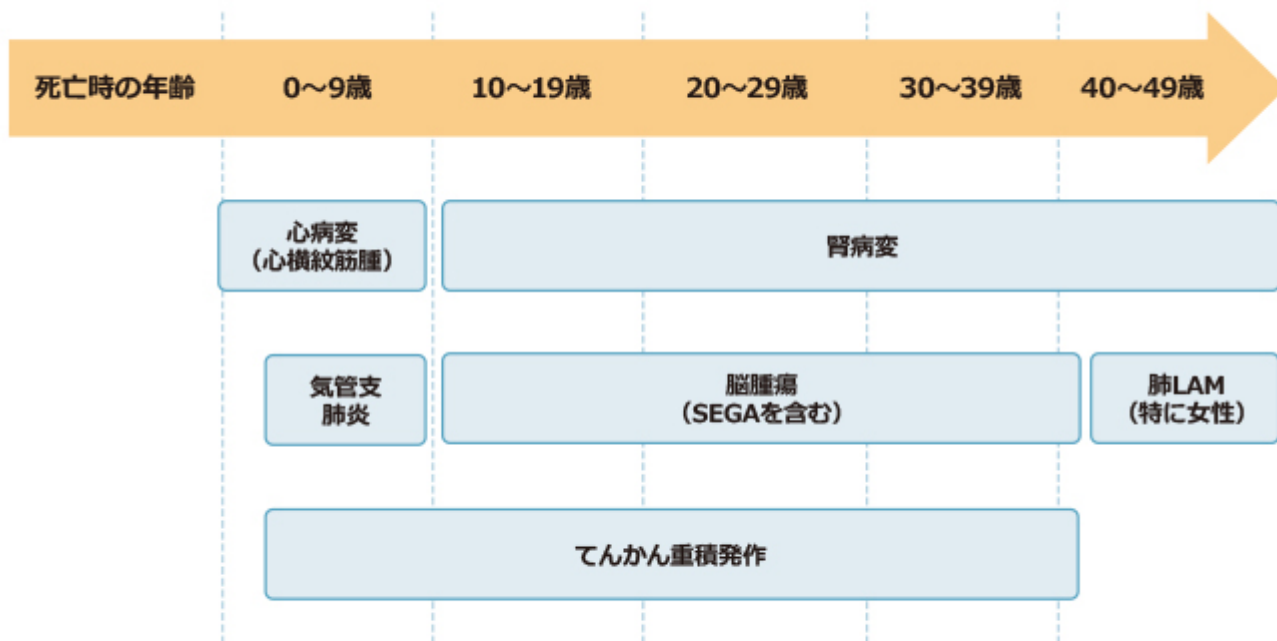
Wataya-Kaneda M, et al. PLoS ONE 2013; 8: e63910

50 80 20 52 130 90 36 10  
 10 19 30 39 1

Image



図2 結節性硬化症の年齢期ごと起こり得る死因



SEGA: 上衣下巨細胞性星細胞腫、肺LAM: 肺リンパ脈管筋腫症

Umeoka S, et al. RadioGraphics 2008; 28: e32より作図

□□□□

- 1) Agata T. Gann Monograph on Cancer Research 1999; 46: 27-35
- 2) Agata T, et al. Annual Report of the Research Committee of Neurocutaneous Syndrome, the Ministry of Health and Welfare 1994: 8-14
- 3) Ahlsén G, et al. Arch Neurol 1994; 51: 76-81
- 4) National Institute of Neurological Disorders and Stroke(NINDS).National Institute of Health(NIH).Tuberous Sclerosis Fact Sheet.2012.
- 5) Osborne JP, et al. Ann N Y Acad Sci 1991; 615: 125-127
- 6) Umeoka S, et al. Radiographics 2008; 28: e32
- 7) Wataya-Kaneda M, et al. PLoS ONE 2013; 8: e63910
- 8) Dabora SL, et al. Am J Hum Genet 2001; 68: 64-80
- 9) Sancak O, et al. Eur J Hum Genet 2005; 13: 731-741.
- 10) Chopra M, et al.J Paediatr Child Health 2011; 47: 711-716.
- 11) Hallett L, et al.Curr Med Res Opin 2011; 27: 1571-1583.
- 12) Shepherd CW, et al. Mayo Clin Proc 1991; 66: 792-796

□□□□□□□□□□□□□□□□

□□□□□□

Source URL: [https://prod.pro.novartis.com/jp-ja/products/afinitor/tsc/overview\\_01](https://prod.pro.novartis.com/jp-ja/products/afinitor/tsc/overview_01)