

## Tuberöse Sklerose: Pathogenese, Klinik und neue Therapieansätze

### Sclérose tubéreuse: pathogénèse, clinique et nouvelles options thérapeutiques

#### Literatur / Références

- 1 Crino PB, Nathanson KL, Henske EP: The tuberous sclerosis complex. *N Engl J Med* 2006;355:(13):1345–1356.
- 2 Osborne JP, Fryer A, Webb D: Epidemiology of tuberous sclerosis. *Ann N Y Acad Sci* 1991;615:125–127.
- 3 Dabora SL, Jozwiak S, Franz DN et al: Mutational analysis in a cohort of 224 tuberous sclerosis patients indicates increased severity of TSC2, compared with TSC1, disease in multiple organs. *Am J Hum Genet* 2001;68:(1):64–80.
- 4 Ess KC, Kamp CA, Tu BP, Gutmann DH: Developmental origin of subependymal giant cell astrocytoma in tuberous sclerosis complex. *Neurology* 2005;64:(8):1446–1449.
- 5 de Vries PJ, Hunt A, Bolton PF: The psychopathologies of children and adolescents with tuberous sclerosis complex (TSC): a postal survey of UK families. *Eur Child Adolesc Psychiatry* 2007;16:(1):16–24.
- 6 Chu-Shore CJ, Major P, Camposano S, Muzykewicz D, Thiele EA: The natural history of epilepsy in tuberous sclerosis complex. *Epilepsia* 2010;51:(7):1236–1241.
- 7 Curatolo P, Jozwiak S, Nababout R: Management of epilepsy associated with tuberous sclerosis complex (TSC): clinical recommendations. *Eur J Paediatr Neurol* 2012;16:(6):582–586.
- 8 Kaufman McNamara E, Curtis AR, Fleischer AB, Jr.: Successful treatment of angiofibromata of tuberous sclerosis complex with rapamycin. *J Dermatolog Treat* 2012;23:(1):46–48.
- 9 Sampson JR, Maheshwar MM, Aspinwall R et al: Renal cystic disease in tuberous sclerosis: role of the polycystic kidney disease 1 gene. *Am J Hum Genet* 1997;61:(4):843–851.
- 10 Yates JR, Maclean C, Higgins JN et al: The Tuberous Sclerosis 2000 Study: presentation, initial assessments and implications for diagnosis and management. *Arch Dis Child* 2011;96:(11):1020–1025.
- 11 O'Callaghan FJ, Clarke AC, Joffe H et al: Tuberous sclerosis complex and Wolff-Parkinson-White syndrome. *Arch Dis Child* 1998;78:(2):159–162.
- 12 Roberds SL: TSC Clinical Consensus Conference Leads to New Diagnosis, Surveillance and Management Recommendations. In: *Perspective*. vol. 129: Tuberous Sclerosis Alliance; 2013: 6–8.
- 13 Braun M, Young J, Reiner CS et al: Ovarian toxicity from sirolimus. *N Engl J Med* 2012;366:(11):1062–1064.
- 14 Augustine JJ, Bodziak KA, Hricik DE: Use of sirolimus in solid organ transplantation. *Drugs* 2007;67:(3):369–391.
- 15 Krueger DA, Care MM, Holland K et al: Everolimus for subependymal giant-cell astrocytomas in tuberous sclerosis. *N Engl J Med* 2010;363:(19):1801–1811.
- 16 Franz DN, Belousova E, Sparagana S et al: Efficacy and safety of everolimus for subependymal giant cell astrocytomas associated with tuberous sclerosis complex (EXIST-1): a multicentre, randomised, placebo-controlled phase 3 trial. *Lancet* 2013;381:(9861):125–132.
- 17 Jozwiak S, Kotulska K, Domanska-Pakiela D et al: Antiepileptic treatment before the onset of seizures reduces epilepsy severity and risk of mental retardation in infants with tuberous sclerosis complex. *Eur J Paediatr Neurol* 2011;15:(5):424–431.
- 18 Bissler JJ, Kingswood JC, Radzikowska E et al: Everolimus for angiomyolipoma associated with tuberous sclerosis complex or sporadic lymphangioleiomyomatosis (EXIST-2): a multicentre, randomised, double-blind, placebo-controlled trial. *Lancet* 2013.
- 19 Bissler JJ, McCormack FX, Young LR et al: Sirolimus for angiomyolipoma in tuberous sclerosis complex or lymphangioleiomyomatosis. *N Engl J Med* 2008;358:(2):140–151.
- 20 Davies DM, Johnson SR, Tattersfield AE et al: Sirolimus therapy in tuberous sclerosis or sporadic lymphangioleiomyomatosis. *N Engl J Med* 2008;358:(2):200–203.
- 21 Schepis C, Siragusa M: Cryosurgery: an easy and cheap therapy for facial angiofibromas in tuberous sclerosis. *Eur J Dermatol* 2010;20:(4):506–507.
- 22 Weiss ET, Geronemus RG: New technique using combined pulsed dye laser and fractional resurfacing for treating facial angiofibromas in tuberous sclerosis. *Lasers Surg Med* 2010;42:(5):357–360.
- 23 Hofbauer GF, Marcollo-Pini A, Corsenca A et al: The mTOR inhibitor rapamycin significantly improves facial angiofibroma lesions in a patient with tuberous sclerosis. *Br J Dermatol* 2008;159:(2):473–475.

- 24 Koenig MK, Hebert AA, Roberson J et al: Topical rapamycin therapy to alleviate the cutaneous manifestations of tuberous sclerosis complex: a double-blind, randomized, controlled trial to evaluate the safety and efficacy of topically applied rapamycin. *Drugs R D* 2012;12:(3):121–126.
- 25 Haemel AK, O'Brian AL, Teng JM: Topical rapamycin: a novel approach to facial angiofibromas in tuberous sclerosis. *Arch Dermatol* 2010;146:(7):715–718.
- 26 McCormack FX, Inoue Y, Moss J et al: Efficacy and safety of sirolimus in lymphangioleiomyomatosis. *N Engl J Med* 2011;364:(17):1595–1606.
- 27 Hyman MH, Whittemore VH: National Institutes of Health consensus conference: tuberous sclerosis complex. *Arch Neurol* 2000;57:(5):662–665.