
BIOGRAPHICAL SKETCH

NAME: Kotulska-Jóźwiak, Katarzyna

POSITION TITLE: Professor and Head of the Department of Neurology and Epileptology, The Children's Memorial Health Institute, Warsaw, Poland

EDUCATION/TRAINING

INSTITUTION AND LOCATION	DEGREE	Completion Date MM/YYYY	FIELD OF STUDY
Medical University of Silesia, Katowice, Poland	MD	06/1997	Medicine
Medical University of Silesia, Katowice, Poland	PhD	01/2002	neuroscience
Medical University of Silesia, Katowice, Poland	habilitation	01/2009	neuroscience
President of Poland	professor	02/2016	medicine
National Centre for Postgradual Medical Education, Warsaw, Poland	specialist	05/2005	neurology
National Centre for Postgradual Medical Education, Warsaw, Poland	specialist	10/2011	pediatric neurology

A. Personal Statement

I am an academic physician-scientist with experience in basic, translational, and clinical science. My clinical and research interests include epileptogenesis and clinical epilepsy studies in the developing brain. I am also involved in research on the rare disorders affecting the nervous system.

B. Positions and Honors

Positions and Employment

2025 – present – member of the Medical Research Agency Council

2015 – present – Head of the Department of Neurology and Epileptology, The Children's Memorial Health Institute, Warsaw, Poland

2014 – 2017 - Deputy Director for Scientific Affairs, The Children's Memorial Health Institute, Warsaw, Poland

2016- present – full professor at The Children's Memorial Health Institute, Warsaw, Poland

2011 – 2015 - Assistant Professor, Department of Research, The Children's Memorial Health Institute, Warsaw, Poland

2009 - 2015 – Assistant Professor, Department of Neurology and Epileptology, The Children's Memorial Health Institute, Warsaw

2010-2011 - Assistant Professor, Department of Clinical and Experimental Neuropathology, Polish Academy of Sciences, Warsaw, Poland

2005-2008 – Assistant Professor of the Department of Neurology and Epileptology, The Children's Memorial Health Institute, Warsaw

2004-2005 – Research Assistant at The Institute of Psychiatry and Neurology, Warsaw

2002-2003 – post-doc at the Universite Paris-7, Paris, France

1997 – 2002 – Research Assistant at the Department of Neurology, Medical University of Silesia, Katowice, Poland

Other Experience and Professional Memberships

2024 - - member, Polish Board for Rare Disorders

2023 - - chair, Ethics Board at The Children's Memorial Health Institute, Warsaw, Poland

2023 - - member of the Medical Advisory Group of the Polish Spinal Muscular Atrophy Foundation

2019 – 2025 - chair of the Board of Heads of the Departments, The Children's Memorial Health Institute

- 2018 – - member, Braincity project external advisory board
- 2018 - - chair of Polish National Spinal Muscular Atrophy Treatment Programme Board
- 2018 – 2020 - vice-president, Polish Board for Rare Disorders
- 2017- - representative of Poland, Board of Member States for Reference Networks, European Commission
- 2016- - faculty member, Polish Mother's Memorial Hospital Research Institute, Łódź, Poland
- 2015- - member of the Medical Advisory Board of the Polish Multiple Sclerosis Association (vice president since 2022)
- 2009 - - faculty member, The Children's Memorial Health Institute, Warsaw, Poland
- 2014 – 2023 - member, Ethics Board at The Children's Memorial Health Institute, Warsaw, Poland
- 2010 - 2011 - Assistant Professor, Department of Clinical and Experimental Neuropathology, Polish Academy of Sciences, Warsaw, Poland
- 2005-2008 – Assistant Professor of the Department of Neurology and Epileptology, The Children's Memorial Health Institute, Warsaw
- 2004-2005 – Assistant at The Institute of Psychiatry and Neurology, Warsaw
- 1997-2006 – research training at the Department of Physiology, Medical University of Silesia, Katowice, Poland

Honors

- 2025 – “Ambassador of Multiple Sclerosis” award
- 2020, 2023, 2024 – List of 100 Most Influential People in Polish Medicine
(<https://pulsmedycyny.pl/medycyna/zdrowie/lista-stu-2019-medycyna/>)
- 2024 – “Women in Medicine in Poland” award
- 2022 – Konorski Award of the Polish Academy of Sciences
- 2016, 2018, 2019, 2022 – Warsaw Medical University Scientific Award
- 2007 – Polish UNESCO and L'Oreal for Women in Science Award
- 2004, 2003, 2002 – Medical University of Silesia Rector's Award
- 2002 – EGIDE fellowship for post-docs

C. Contribution to Science

1. Regeneration in the optic nerve and peripheral nerves (1997–2006)

In contrast to peripheral nerves, central nervous system neurons do not show the potential for spontaneous repair. My earliest research interest focused on this difference. I tried to induce the regeneration of central neurons by mimicking the milieu of peripheral nerves in animal models. I have also investigated the phenomenon of neuropathic pain after peripheral nerve injury. Our team proposed the innovative technique of peripheral nerve repair to prevent the development of neuroma.

- a. Marcol W, **Kotulska K**, Swiech-Sabuda E, Larysz-Brysz M, Gołka B, Górka D, Lewin-Kowalik J. Regeneration of sciatic nerves of adult rats induced by extracts from distal stumps of pre-degenerated peripheral nerves. *J Neurosci Res.* 2003 May 1;72(3):417-24.
- b. **Kotulska K**, Lewin-Kowalik J, Jaroslaw-Jerzy B, Larysz-Brysz M, Marcol W, Fus Z. Bcl-2 deficiency deprives peripheral nerves of neurotrophic activity against injured optic nerve. *J Neurosci Res.* 2003 Sep 15;73(6):846-52. Erratum in: *J Neurosci Res.* 2004 Jul 1;77(1):153.
- c. **Kotulska K**, Marcol W, Larysz-Brysz M, Tendera Z, Malinowska-Kołodziej I, Słusarczyk W, Jedzejewska-Szypułka H, Lewin-Kowalik J. Effect of oblique nerve grafting on peripheral nerve regeneration in rats. *Microsurgery.* 2006;26(8):579-84.
- d. **Kotulska K**, Larysz-Brysz M, Marcol W, Grajkowska W, Józwiak S, Lewin-Kowalik J. The role of trkB receptor in the formation of post-traumatic neuroma. *Folia Neuropathol.* 2006;44(3):221-7.
- e. **Kotulska K**, LePecheur M, Marcol W, Lewin-Kowalik J, Larysz-Brysz M, Paly E, Matuszek I, London J. Overexpression of copper/zinc-superoxide dismutase in transgenic mice markedly impairs regeneration and increases development of neuropathic pain after sciatic nerve injury. *J Neurosci Res.* 2006 Oct;84(5):1091-7.
- f. **Kotulska K**, Marcol W, Larysz-Brysz M, Barski JJ, Fus Z, Lewin-Kowalik J. Impaired regeneration of bcl-2-lacking peripheral nerves. *Neurol Res.* 2005 Dec;27(8):843-9.

2. Development of tumors in TSC (2002–present)

In TSC, non-malignant tumors develop in the brain, skin, kidney, liver, retina and other organs over time. My research focused on neuropathology of TSC and the clinical presentation of the disease in children, and particularly newborns and infants.

- a. Klonowska K, Giannikou K, Grevelink JM, Boeszoermerenyi B, Thorner AR, Herbert ZT, Afrin A, Treichel AM, Hamieh L, **Kotulska K**, Jozwiak S, Moss J, Darling TN, Kwiatkowski DJ. Comprehensive genetic and phenotype analysis of 95 individuals with mosaic tuberous sclerosis complex. *Am J Hum Genet.* 2023 Jun 1;110(6):979-988.
- b. Gao C, Zabielska B, Jiao F, Mei D, Wang X, **Kotulska K**, Jozwiak S. Subependymal Giant Cell Astrocytomas in Tuberous Sclerosis Complex-Current Views on Their Pathogenesis and Management. *J Clin Med.* 2023 Jan 26;12(3):956. doi: 10.3390/jcm12030956.
- c. Klonowska K, Giannikou K, Grevelink JM, Boeszoermerenyi B, Thorner AR, Herbert ZT, Afrin A, Treichel AM, Hamieh L, **Kotulska K**, Jozwiak S, Moss J, Darling TN, Kwiatkowski DJ. Comprehensive genetic and phenotype analysis of 95 individuals with mosaic tuberous sclerosis complex. *Am J Hum Genet.* 2023 Apr 28:S0002-9297(23)00126-X. doi: 10.1016/j.ajhg.2023.04.002. Online ahead of print.
- d. Bongaarts A, Mijnsbergen C, Anink JJ, Jansen FE, Spliet WGM, den Dunnen WFA, Coras R, Blümcke I, Paulus W, Gruber VE, Scholl T, Hainfellner JA, Feucht M, **Kotulska K**, Jozwiak S, Grajkowska W, Buccoliero AM, Caporalini C, Giordano F, Genitori L, Söylemezoğlu F, Pimentel J, Jones DTW, Scicluna BP, Schouten-van Meeteren AYN, Mühlebner A, Mills JD, Aronica E Distinct DNA Methylation Patterns of Subependymal Giant Cell Astrocytomas in Tuberous Sclerosis Complex. *Cell Mol Neurobiol.* 2021 Oct 28. doi: 10.1007/s10571-021-01157-5.
- e. Stępnia K, Machnicka MA, Mieczkowski J, Macioszek A, Wojtaś B, Gielniewski B, Poleszak K, Perycz M, Król SK, Guzik R, Dąbrowski MJ, Dramiński M, Jardańska M, Grabowicz I, Dziedzic A, Kranas H, Sienkiewicz K, Diamanti K, Kotulska K, Grajkowska W, Roszkowski M, Czernicki T, Marchel A, Komorowski J, Kaminska B, Wilczyński B. Mapping chromatin accessibility and active regulatory elements reveals pathological mechanisms in human gliomas. *Nat Commun.* 2021 Jun 15;12(1):3621. doi: 10.1038/s41467-021-23922-2.
- f. Koscielny A, Liszewska E, Machnicka K, Wezyk M, Kotulska K, Jaworski J. mTOR controls endoplasmic reticulum-Golgi apparatus trafficking of VSVg in specific cell types. *Cell Mol Biol Lett.* 2021 May 18;26(1):18. doi: 10.1186/s11658-021-00262-z.
- g. Korotkov A, Luinenburg MJ, Romagnolo A, Zimmer TS, van Scheppingen J, Bongaarts A, Broekaart DWM, Anink JJ, Mijnsbergen C, Jansen FE, van Hecke W, Spliet WG, van Rijen PC, Feucht M, Hainfellner JA, Krsek P, Zamecnik J, Crino PB, **Kotulska K**, Lagae L, Jansen AC, Kwiatkowski DJ, Jozwiak S, Curatolo P, Mühlebner A, van Vliet EA, Mills JD, Aronica E. Down-regulation of the brain-specific cell-adhesion molecule contactin-3 in tuberous sclerosis complex during the early postnatal period. *J Neurodev Disord.* 2022 Jan 15;14(1):8. doi: 10.1186/s11689-022-09416-2.
- h. Koscielny A, Liszewska E, Machnicka K, Wezyk M, Kotulska K, Jaworski J. mTOR controls endoplasmic reticulum-Golgi apparatus trafficking of VSVg in specific cell types. *Cell Mol Biol Lett.* 2021 May 18;26(1):18. doi: 10.1186/s11658-021-00262-z.
- i. Liszewska E, Majchrowicz L, Krogulec E, Kotulska K, Kaczmarek L, Kalita K, Dobrzyń A, Jaworski J. Establishment of two hiPSC lines (IIMCBI001-A and IIMCBI002-A) from dermal fibroblasts of healthy donors and characterization of their cell cycle. *Stem Cell Res.* 2021 Apr;52:102225. doi: 10.1016/j.scr.2021.102225. Epub 2021 Feb 5.
- j. Bąbol-Pokora K, Bielska M, Bobeff K, Jatczak-Pawlik I, Borkowska J, **Kotulska K**, Józwiak S, Młynarski W, Trelńska J. A multistep approach to the genotype-phenotype analysis of polish patients with tuberous sclerosis complex. *Eur J Med Genet.* 2021 Aug 14:104309. doi: 10.1016/j.ejmg.2021.104309. Online ahead of print.
- k. Giannikou K, Zhu Z, Kim J, Winden KD, Tyburczy ME, Marron D, Parker JS, Hebert Z, Bongaarts A, Taing L, Long HW, Pisano WV, Alexandrescu S, Godlewski B, Nellist M, **Kotulska K**, Jozwiak S, Roszkowski M, Mander M, Thiele EA, Lidov H, Getz G, Devinsky O, Lawrence MS, Ligon KL, Ellison DW, Sahin M, Aronica E, Meredith DM, Kwiatkowski DJ. Subependymal giant cell astrocytomas are characterized by mTORC1 hyperactivation, a very low somatic mutation rate, and a unique gene expression profile. *Mod Pathol.* 2021 Feb;34(2):264-279. doi: 10.1038/s41379-020-00659-9.
- l. Ogórek B, Hamieh L, Hulshof HM, Lasseter K, Klonowska K, Kuijff H, Moavero R, Hertzberg C, Weschke B, Riney K, Feucht M, Scholl T, Krsek P, Nabbout R, Jansen AC, Benova B, Aronica E, Lagae L, Curatolo P, Borkowska J, Sadowski K, Domańska-Pakieła D, Janson S, Kozłowski P,

Urbanska M, Jaworski J, Jozwiak S, Jansen FE, **Kotulska K**; EPISTOP Consortium members, Kwiatkowski DJ. TSC2 pathogenic variants are predictive of severe clinical manifestations in TSC infants: results of the EPISTOP study. *Genet Med.* 2020 Sep;22(9):1489-1497. doi: 10.1038/s41436-020-0823-4. Epub 2020 May 28.

- m. Bongaarts A, de Jong JM, Broekaart DWM, van Scheppingen J, Anink JJ, Mijnsbergen C, Jansen FE, Spliet WGM, den Dunnen WFA, Gruber VE, Scholl T, Hainfellner JA, Feucht M, Borkowska J, **Kotulska K**, Jozwiak S, Grajkowska W, Buccoliero AM, Caporalini C, Giordano F, Genitori L, Scicluna BP, Schouten-van Meeteren AYN, van Vliet EA, Mühlebner A, Mills JD, Aronica E. Dysregulation of the MMP/TIMP Proteolytic System in Subependymal Giant Cell Astrocytomas in Patients With Tuberous Sclerosis Complex: Modulation of MMP by MicroRNA-320d In Vitro. *J Neuropathol Exp Neurol.* 2020 Jul 1;79(7):777-790. doi: 10.1093/jnen/nlaa040.
- n. Mühlebner A, van Scheppingen J, de Neef A, Bongaarts A, Zimmer TS, Mills JD, Jansen FE, Spliet WGM, Krsek P, Zamecnik J, Coras R, Blumcke I, Feucht M, Scholl T, Gruber VE, Hainfellner JA, Söylemezoğlu F, **Kotulska K**, Lagae L, Jansen AC, Kwiatkowski DJ, Jozwiak S, Curatolo P, Aronica E. Myelin Pathology Beyond White Matter in Tuberous Sclerosis Complex (TSC) Cortical Tubers. *J Neuropathol Exp Neurol.* 2020 Oct 1;79(10):1054-1064. doi: 10.1093/jnen/nlaa090.
- o. Bongaarts A, van Scheppingen J, Korotkov A, Mijnsbergen C, Anink JJ, Jansen FE, Spliet WGM, den Dunnen WFA, Gruber VE, Scholl T, Samuelli S, Hainfellner JA, Feucht M, **Kotulska K**, Jozwiak S, Grajkowska W, Buccoliero AM, Caporalini C, Giordano F, Genitori L, Coras R, Blümcke I, Krsek P, Zamecnik J, Meijer L, Scicluna BP, Schouten-van Meeteren AYN, Mühlebner A, Mills JD, Aronica E. The coding and non-coding transcriptional landscape of subependymal giant cell astrocytomas. *Brain.* 2020 Jan 1;143(1):131-149.
- p. Urbanska M, Kazmierska-Grebowska P, Kowalczyk T, Caban B, Nader K, Pijet B, Kalita K, Gozdz A, Devijver H, Lechat B, Jaworski T, Grajkowska W, Sadowski K, Jozwiak S, **Kotulska K**, Konopacki J, Van Leuven F, van Vliet EA, Aronica E, Jaworski J. GSK3 β activity alleviates epileptogenesis and limits GluA1 phosphorylation. *EBioMedicine.* 2019 Jan;39:377-387.
- q. Józwiak S, Sadowski K, Borkowska J, Domańska-Pakieła D, Chmielewski D, Jurkiewicz E, Jaworski M, Urbańska M, Ogrodnik M, Słowińska M, **Kotulska K**. Liver Angiomyolipomas in Tuberous Sclerosis Complex-Their Incidence and Course. *Pediatr Neurol.* 2018 Jan;78:20-26.
- r. Kuzniewska B, Sadowski K, Urbanska K, Urbanska M, **Kotulska K**, Liszewska E, Grajkowska W, Józwiak S, Dziembowska M. The level of microRNA 21 is upregulated by rapamycin in serum of tuberous sclerosis complex patients and subependymal giant cell astrocytoma (SEGA)-derived cell cultures. *Folia Neuropathol.* 2018;56(3):167-174.
- s. Bongaarts A, Prabowo AS, Arena A, Anink JJ, Reinten RJ, Jansen FE, Spliet WGM, Thom M, Coras R, Blümcke I, **Kotulska K**, Jozwiak S, Grajkowska W, Söylemezoğlu F, Pimentel J, Schouten-van Meeteren AYN, Mills JD, Iyer AM, van Vliet EA, Mühlebner A, Aronica E. MicroRNA519d and microRNA4758 can identify gangliogliomas from dysembryoplastic neuroepithelial tumours and astrocytomas. *Oncotarget.* 2018 Jun 15;9(46):28103-28115.
- t. Słowińska M, Józwiak S, Peron A, Borkowska J, Chmielewski D, Sadowski K, Jurkiewicz E, Vignoli A, La Briola F, Canevini MP, **Kotulska-Józwiak K**. Early diagnosis of tuberous sclerosis complex: a race against time. How to make the diagnosis before seizures? *Orphanet J Rare Dis.* 2018 Jan 29;13(1):25.
- u. Bongaarts A, Giannikou K, Reinten RJ, Anink JJ, Mills JD, Jansen FE, Spliet GMW, den Dunnen WFA, Coras R, Blümcke I, Paulus W, Scholl T, Feucht M, **Kotulska K**, Jozwiak S, Buccoliero AM, Caporalini C, Giordano F, Genitori L, Söylemezoğlu F, Pimentel J, Nellist M, Schouten-van Meeteren AYN, Nag A, Mühlebner A, Kwiatkowski DJ, Aronica E. Subependymal giant cell astrocytomas in Tuberous Sclerosis Complex have consistent TSC1/TSC2biallelic inactivation, and no BRAF mutations. *Oncotarget.* 2017 Sep 8;8(56):95516-95529.
- v. Stawiski K, Trelińska J, Baranska D, Dachowska I, **Kotulska K**, Józwiak S, Fendler W, Młynarski W. What are the true volumes of SEGA tumors? Reliability of planimetric and popular semi-automated image segmentation methods. *MAGMA.* 2017 Aug;30(4):397-405.
- w. van Scheppingen J, Iyer AM, Prabowo AS, Mühlebner A, Anink JJ, Scholl T, Feucht M, Jansen FE, Spliet WG, Krsek P, Zamecnik J, Buccoliero AM, Giordano F, Genitori L, **Kotulska K**, Jozwiak S, Jaworski J, Liszewska E, van Vliet EA, Aronica E. Expression of microRNAs miR21, miR146a, and miR155 in tuberous sclerosis complex cortical tubers and their regulation in human astrocytes and SEGA-derived cell cultures. *Glia.* 2016 Jun;64(6):1066-82.

- x. Malik AR, Liszewska E, Skalecka A, Urbanska M, Iyer AM, Swiech LJ, Perycz M, Parobczak K, Pietruszka P, Zarebska MM, Macias M, **Kotulska K**, Borkowska J, Grajkowska W, Tyburczy ME, Jozwiak S, Kwiatkowski DJ, Aronica E, Jaworski J. Tuberous sclerosis complex neuropathology requires glutamate-cysteine ligase. *Acta Neuropathol Commun*. 2015 Jul 30;3:48.
- y. **Kotulska K**, Borkowska J, Mandra M, Roszkowski M, Jurkiewicz E, Grajkowska W, Bilka M, Jóźwiak S. Congenital subependymal giant cell astrocytomas in patients with tuberous sclerosis complex. *Childs Nerv Syst*. 2014 Dec;30(12):2037-42.
- z. **Kotulska K**, Larysz-Brysz M, Grajkowska W, Jóźwiak J, Włodarski P, Sahin M, Lewin-Kowalik J, Domańska-Pakieła D, Jóźwiak S. Cardiac rhabdomyomas in tuberous sclerosis complex show apoptosis regulation and mTOR pathway abnormalities. *Pediatr Dev Pathol*. 2009 Mar-Apr;12(2):89-95.
- aa. Tyburczy ME, **Kotulska K**, Pokarowski P, Mieczkowski J, Kucharska J, Grajkowska W, Roszkowski M, Jozwiak S, Kaminska B. Novel proteins regulated by mTOR in subependymal giant cell astrocytomas of patients with tuberous sclerosis complex and new therapeutic implications. *Am J Pathol*. 2010 Apr;176(4):1878-90.
- bb. Jozwiak J, **Kotulska K**, Grajkowska W, Jozwiak S, Zalewski W, Oldak M, Lojek M, Rainko K, Maksym R, Lazarczyk M, Skopinski P, Wlodarski P. Upregulation of the WNT pathway in tuberous sclerosis-associated subependymal giant cell astrocytomas. *Brain Dev*. 2007 Jun;29(5):273-80.
- cc. Jozwiak J, **Kotulska K**, Jozwiak S. Similarity of balloon cells in focal cortical dysplasia to giant cells in tuberous sclerosis. *Epilepsia*. 2006 Apr;47(4):805.
- dd. Jóźwiak S, Kwiatkowski D, **Kotulska K**, Larysz-Brysz M, Lewin-Kowalik J, Grajkowska W, Roszkowski M. Tuberin and hamartin expression is reduced in the majority of subependymal giant cell astrocytomas in tuberous sclerosis complex consistent with a two-hit model of pathogenesis. *J Child Neurol*. 2004 Feb;19(2):102-6.
- ee. Chan JA, Zhang H, Roberts PS, Jozwiak S, Wiesława G, Lewin-Kowalik J, **Kotulska K**, Kwiatkowski DJ. Pathogenesis of tuberous sclerosis subependymal giant cell astrocytomas: biallelic inactivation of TSC1 or TSC2 leads to mTOR activation. *J Neuropathol Exp Neurol*. 2004 Dec;63(12):1236-42.

3. Epileptogenesis in TSC (2003–present)

Epilepsy affects about 80-90% of TSC patients and in most cases develops in infancy. It is frequently associated with intellectual disability and autistic features. Our studies proved that seizures in TSC are preceded by latent epileptogenesis which may be captured by EEG recording. Treatment in this latent period reduces the risk of seizures, the risk of drug-resistant epilepsy and intellectual disability in TSC. We have proposed such approach in international, multicenter project called EPISTOP. We investigate it further in another project called EPIMARKER. In 2020, we launched a non-commercial, randomized clinical trial comparing the efficacy and safety of rapamycin vs vigabatrin as a preventive treatment in TSC infants (ViRAP)

- a. Jóźwiak S, Curatolo P, **Kotulska K**. Intellectual disability and autistic behavior and their modifying factors in children with tuberous sclerosis complex. *Brain Dev*. 2025 Jan 31;47(2):104322. doi: 10.1016/j.braindev.2025.104322. Epub ahead of print. PMID: 39891979.
- b. Huschner F, Głowacka-Walas J, Mills JD, Klonowska K, Lasseter K, Asara JM, Moavero R, Hertzberg C, Weschke B, Riney K, Feucht M, Scholl T, Krsek P, Nabbout R, Jansen AC, Petrák B, van Scheppingen J, Zamecnik J, Iyer A, Anink JJ, Mühlebner A, Mijnsbergen C, Lagae L, Curatolo P, Borkowska J, Sadowski K, Domańska-Pakieła D, Blazejczyk M, Jansen FE, Janson S, Urbanska M, Tempes A, Janssen B, Sijko K, Wojdan K, Jozwiak S, **Kotulska K**, Lehmann K, Aronica E, Jaworski J, Kwiatkowski DJ. Molecular EPISTOP, a comprehensive multi-omic analysis of blood from Tuberous Sclerosis Complex infants age birth to two years. *Nat Commun*. 2023 Nov 23;14(1):7664. doi: 10.1038/s41467-023-42855-6.
- c. Foryś-Basiejko M, **Kotulska K**, Maryniak A, Siłuszyk A, Szkop M, Borkowska J, Sugalska M, Głowacka-Walas J, Jóźwiak S. Epilepsy and Language Development in 8-36-Month-Old Toddlers with Tuberous Sclerosis Complex. *J Clin Med*. 2022 Aug 4;11(15):4564. doi: 10.3390/jcm11154564.
- d. Scheper M, Romagnolo A, Besharat ZM, Iyer AM, Moavero R, Hertzberg C, Weschke B, Riney K, Feucht M, Scholl T, Petrak B, Maulisova A, Nabbout R, Jansen AC, Jansen FE, Lagae L, Urbanska M, Ferretti E, Tempes A, Blazejczyk M, Jaworski J, Kwiatkowski DJ, Jozwiak S, **Kotulska K**, Sadowski K,

Borkowska J, Curatolo P, Mills JD, Aronica E, Epistop Consortium Members. miRNAs and isomiRs: Serum-Based Biomarkers for the Development of Intellectual Disability and Autism Spectrum Disorder in Tuberous Sclerosis Complex. *Biomedicines*. 2022 Jul 29;10(8):1838. doi: 10.3390/biomedicines10081838.

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- f. De Ridder J, **Kotulska K**, Curatolo P, Jansen AC, Aronica E, Kwiatkowski DJ, Jansen FE, Jóźwiak S, Lagae L; Epistop Consortium. Evolution of electroencephalogram in infants with tuberous sclerosis complex and neurodevelopmental outcome: a prospective cohort study. *Dev Med Child Neurol*. 2022 Apr;64(4):495-501. doi: 10.1111/dmcn.15073. Epub 2021 Oct 2.
- g. Korotkov A, Sim NS, Luinenburg MJ, Anink JJ, van Scheppingen J, Zimmer TS, Bongaarts A, Broekaart DWM, Mijnsbergen C, Jansen FE, Van Hecke W, Spliet WGM, van Rijen PC, Feucht M, Hainfellner JA, Kršek P, Zamecnik J, Crino PB, Kotulska K, Lagae L, Jansen AC, Kwiatkowski DJ, Jozwiak S, Curatolo P, Mühlebner A, Lee JH, Mills JD, van Vliet EA, Aronica E. MicroRNA-34a activation in tuberous sclerosis complex during early brain development may lead to impaired corticogenesis. *Neuropathol Appl Neurobiol*. 2021 May 3. doi: 10.1111/nan.12717. Online ahead of print.
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4. Treatment in TSC (2007–present)

Abnormal activation of mTOR pathway is a hallmark of TSC. Rapamycin and its analog, everolimus, have been introduced in the treatment of TSC and its symptoms: SEGA, AMLs, epilepsy and facial angiofibroma. My research focuses on the safety and efficacy of mTOR inhibition in TSC. I have also proposed that mTOR inhibition might prevent the development of tumors in TSC. This concept is addressed in the ViRap study, which aims to compare the safety and efficacy of mTOR inhibitor (Rapamycin) and antiepileptic drug (vigabatrin) in preventive treatment of TSC infants. I am also coordinating a non-commercial clinical trial RaRE-TS, evaluating the efficacy and safety of rapamycin vs placebo in drug-resistant epilepsy associated with TSC.

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Complete List of Published Work from PubMed (occasional publications not captured due to name misspelling):

<https://www.ncbi.nlm.nih.gov/pubmed/?term=kotulska+k+or++kotulska-jozwiak>

D. Additional Information: Research Support and/or Scholastic Performance Selected Research Support

BraimTOR (PI: Trubicka)

01/09/21 – 31/08/27

Polish National Medical Research Agency (ABM)

An open randomized phase II clinical trial evaluating the safety and efficacy of rapamycin in the treatment of drug-resistant epilepsy in children as part of the establishment management of rare and ultra rare diseases of the central nervous system associated with mTOR pathway activation

The major goals of this project are: 1) to carry out a randomized open-label clinical trial of rapamycin as an add-on treatment in children with drug-resistant epilepsy related to mTOR pathway activation; 2) to identify the biomarkers of drug-resistant epilepsy related to mTOR pathway abnormalities.

Role: workpackage leader

Rare-TS (PI: Kotulska)

01/01/21 – 12/31/26

Polish National Medical Research Agency (ABM)

Multicenter, randomized, double-blind, placebo controlled, phase III study to assess the efficacy and safety of RArapamycin in drug Resistant Epilepsy associated with TSC (RaRE-TS)

The major goals of this project are: 1) to carry out a randomized placebo-controlled clinical trial of rapamycin as an add-on treatment in children with drug-resistant epilepsy related to TSC; 2) to identify the biomarkers of drug-resistant epilepsy in patients with TSC.

Role: Principal Investigator

VIRAP (PI: Kotulska)

04/01/20 – 3/31/27

Polish National Medical Research Agency (ABM)

Multicenter, randomized, double-blind and double-dummy clinical trial comparing the safety, tolerability, and efficacy of vigabatrin and rapamycin in preventive treatment of infants with Tuberous Sclerosis Complex

The major goals of this project are: 1) to carry out a randomized clinical trial of vigabatrin versus rapamycin in preventive treatment in children with TSC; 2) to identify the biomarkers of epileptogenesis and tumorigenesis in children with TSC.

Role: Principal Investigator

EPIMARKER (PI: Jozwiak – Co-Inv: Kotulska)

04/01/16 – 3/31/20

Polish National for Research and Development (STRATEGMED program)

Application of Novel Diagnostic and Therapeutic Methods in Epilepsy and Neurodevelopmental Abnormalities in Children.

The major goals of this project are: 1) to carry out a prospective study on the development of drug-resistant epilepsy in children with TSC; 2) to evaluate the risk of epilepsy recurrence in children with TSC, in whom antiepileptic drugs were withdrawn; 3) to test epileptogenesis in a human-derived organoid model of TSC; 4) produce an integrative tool useful in everyday diagnosis and treatment of epilepsy in children to prevent the development of drug-resistant epilepsy and its behavioral comorbidities.

Role: Co-Investigator and Leader of the Workpackage

European Commission 602391-2 (PI – Jozwiak)

11/01/13 – 04/30/19

EPISTOP: Long-term, prospective study evaluating clinical and molecular biomarkers of epileptogenesis in a genetic model of epilepsy – tuberous sclerosis complex

The goal in WP6 was to perform a multicenter, randomized and open-label study to compare the epilepsy outcome in TSC infants receiving preventive or conventional antiepileptic treatment.

Role: co-Investigator and Leader of WP6