



**Susana Pinto**

**Kontakt**

Susana Pinto

## Publikationen (10)

Adey B, Cooper-Knock J, Al Khleifat A, Fogh I, Van Damme P, Corcia P, Couratier P, Hardiman O, McLaughlin R, Gotkine M, Drory V, Silani V, Ticozzi N, Veldink J, van den Berg L, de Carvalho M, Pinto S, Mora J, Povedano Panades M, Andersen P, Weber M, Başak N, Shaw C, Openshaw P, Morrison K, Landers J, Glass J, Vourc'h P, Dobson R, Breen G, Al-Chalabi A, Jones A, Iacoangeli A. Large-scale analyses of CAV1 and CAV2 suggest their expression is higher in post-mortem ALS brain tissue and affects survival. *Front Cell Neurosci* 2023; 17:1112405.

van Rheenen W, van der Spek R, Bakker M, Van Vugt J, Hop P, Zwamborn R, de Klein N, Westra H, Bakker O, Deelen P, Shireby G, Hannon E, Moisse M, Baird D, Restuadi R, Dolzhenko E, Dekker A, Gawor K, Westeneng H, Tazelaar G, van Eijk K, Kooyman M, Byrne R, Doherty M, Heverin M, Al Khleifat A, Iacoangeli A, Shatunov A, Ticozzi N, Cooper-Knock J, Smith B, Gromicho M, Chandran S, Pal S, Morrison K, Shaw P, Hardy J, Orrell R, Sendtner M, Meyer T, Başak N, van der Kooi A, Ratti A, Fogh I, Gellera C, Lauria G, Corti S, Cereda C, Sproviero D, D'Alfonso S, Sorarù G, Siciliano G, Filosto M, Padovani A, Chio A, Calvo A, Moglia C, Brunetti M, Canosa A, Grassano M, Beghi E, Pupillo E, Logroscino G, Nefussy B, Osmanovic A, Nordin A, Lerner Y, Zabari M, Gotkine M, Baloh R, Bell S, Vourc'h P, Corcia P, Couratier P, Millecamps S, Meininger V, Salachas F, Mora Pardina J, Assialioui A, Rojas-García R, Dion P, Ross J, Ludolph A, Weishaupt J, Brenner D, Freischmidt A, Bensimon G, Brice A, Dürr A, Payan C, Saker-Delye S, Wood N, Topp S, Rademakers R, Tittmann L, Lieb W, Franke A, Ripke S, Braun A, Kraft J, Whiteman D, Olsen C, Uitterlinden A, Hofman A, Rietschel M, Cichon S, Nöthen M, Amouyel P, SLALOM Consortium, PARALS Consortium, SLAGEN Consortium, SLAP Consortium, Traynor B, Singleton A, Mitne Neto M, Cauchi R, Ophoff R, Wiedau-Pazos M, Lomen-Hoerth C, Van Deerlin V, Grosskreutz J, Roediger A, Gaur N, Jörk A, Barthel T, Theele E, Ilse B, Stubendorff B, Witte O, Steinbach R, Hübner C, Graff C, Brylev L, Fominykh V, Demeshonok V, Ataulina A, Rogelj B, Koritnik B, Zidar J, Ravnik-Glavač M, Glavač D, Stević Z, Drory V, Povedano M, Blair I, Kiernan M, Benjamin B, Henderson R, Furlong S, Mathers S, McCombe P, Needham M, Ngo S, Nicholson G, Pamphlett R, Rowe D, Steyn F, Williams K, Mather K, Sachdev P, Henders A, Wallace L, de Carvalho M, Pinto S, Petri S, Weber M, Rouleau G, Silani V, Curtis C, Breen G, Glass J, Brown R, Landers J, Shaw C, Andersen P, Groen E, van Es M, Pasterkamp R, Fan D, Garton F, McRae A, Davey Smith G, Gaunt T, Eberle M, Mill J, McLaughlin R, Hardiman O, Kenna K, Wray N, Tsai E, Runz H, Franke L, Al-Chalabi A, Van Damme P, van den Berg L, Veldink J. Author Correction: Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. *Nat Genet* 2022; 54:361.

Hop P, Zwamborn R, Hannon E, Shireby G, Nabais M, Walker E, van Rheenen W, Van Vugt J, Dekker A, Westeneng H, Tazelaar G, van Eijk K, Moisse M, Baird D, Al Khleifat A, Iacoangeli A, Ticozzi N, Ratti A, Cooper-Knock J, Morrison K, Shaw P, Basak A, Chio A, Calvo A, Moglia C, Canosa A, Brunetti M, Grassano M, Gotkine M, Lerner Y, Zabari M, Vourc'h P, Corcia P, Couratier P, Mora Pardina J, Salas T, Dion P, Ross J, Henderson R, Mathers S, McCombe P, Needham M, Nicholson G, Rowe D, Pamphlett R, Mather K, Sachdev P, Furlong S, Garton F, Henders A, Lin T, Ngo S, Steyn F, Wallace L, Williams K, Neto M, Cauchi R, Blair I, Kiernan M, Drory V, Povedano M, de Carvalho M, Pinto S, Weber M, Rouleau G, Silani V, Landers J, Shaw C, Andersen P, McRae A, van Es M, Pasterkamp R, Wray N, McLaughlin R, Hardiman O, Kenna K, Tsai E, Runz H, Al-Chalabi A, van den Berg L, Van Damme P, Mill J, Veldink J. Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. *Sci Transl Med* 2022; 14:eabj0264.

van Rheenen W, van der Spek R, Bakker M, Farei-Campagna J, Hop P, Zwamborn R, de Klein N, Westra H, Bakker O, Deelen P, Shireby G, Hannon E, Moisse M, Baird D, Restuadi R, Dolzhenko E, Dekker A, Gawor K, Westeneng H, Tazelaar G, van Eijk K, Kooyman M, Byrne R, Doherty M, Heverin M, Al Khleifat A, Iacoangeli A, Shatunov A, Ticozzi N, Cooper-Knock J, Smith B, Gromicho M, Chandran S, Pal S, Morrison K, Openshaw P, Hardy J, Orrell R, Sendtner M, Meyer T, Başak N, van der Kooi A, Ratti A, Fogh I, Gellera C, Lauria G, Corti S, Cereda C, Sproviero D, D'Alfonso S, Sorarù G, Siciliano G, Filosto M, Padovani A, Chio A, Calvo A, Moglia C, Brunetti M, Canosa A, Grassano M, Beghi E, Pupillo E, Logroscino G, Nefussy B, Osmanovic A, Nordin A, Lerner Y, Zabari M, Gotkine M, Baloh R, Bell S, Vourc'h P, Corcia P, Couratier P, Millecamps S, Meininger V, Salachas F, Mora J, Assialioui A, Rojas-García R, Dion P, Ross J, Ludolph A, Weishaupt J, Brenner D, Freischmidt A, Bensimon G, Brice A, Dürr A, Payan C, Saker-Delye S, Wood N, Topp S, Rademakers R, Tittmann L, Lieb W, Klein-Franke A, Ripke S, Braun A, Kraft J, Whiteman D, Olsen C, Uitterlinden A, Hofman A, Rietschel M, Cichon S, Nöthen M, Amouyel P, SLALOM Consortium, PARALS Consortium, SLAGEN Consortium, SLAP Consortium, Traynor B, Singleton A, Mitne Neto M, Cauchi R, Ophoff R, Wiedau-Pazos M, Lomen-Hoerth C, Van Deerlin V, Grosskreutz J, Roediger A, Gaur N, Jörk A, Barthel T, Theele E, Ilse B, Stubendorff B, Witte O, Steinbach R, Hübner C, Graff C, Brylev L, Fominykh V, Demeshonok V, Ataulina A, Rogelj B, Koritnik B, Zidar J, Ravnik-Glavač M, Glavač D, Stević Z, Drory V, Povedano M, Blair I, Kiernan M, Benyamin B, Henderson R, Furlong S, Mathers S, McCombe P, Needham M, Ngo S, Nicholson G, Pamphlett R, Rowe D, Steyn F, Williams K, Mather K, Sachdev P, Henders A, Wallace L, de Carvalho M, Pinto S, Petri S, Weber M, Rouleau G, Silani V, Curtis C, Breen G, Glass J, Brown R, Landers J, Shaw C, Andersen P, Groen E, van Es M, Pasterkamp R, Fan D, Garton F, McRae A, Davey Smith G, Gaunt T, Eberle M, Mill J, McLaughlin R, Hardiman O, Kenna K, Wray N, Tsai E, Runz H, Franke L, Al-Chalabi A, Van Damme P, van den Berg L, Veldink J. Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. *Nat Genet* 2021; 53:1636-1648.

German ALS network MND-NET, Weis J, Dorst J, Volk A, Borck G, Sperfeld A, de Carvalho M, Klopstock T, Sendtner M, Otto M, Schuster J, Weishaupt J, Andersen P, Ludolph A, Meitinger T, Freischmidt A, Danzer K, Braak H, Del Tredici K, Jablonka S, Kubisch C, Zeller D, Weydt P, Grosskreutz J, Meyer T, Petri S, Grehl T, Müller K, Yilmaz R, Rosenbohm A, Ruf W, Neuwirth C, Hübers A, Günther K, Knehr A, Jordan B, Schrank B, Claeys K, Pinto S, Weber M, Brenner D. Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. *Brain* 2019; 142:e67.

van den Berg L, Grosskreutz J, Weber M, Couratier P, Corcia P, van Es M, Bredenoord A, Shaw C, Gunkel A, Rödiger A, Ludolph A, Petri S, de Carvalho M, Veldink J, Moons K, Hardiman O, Chio A, Al-Chalabi A, Shaw P, Turner M, Talbot K, Van Damme P, Ringer T, Körner S, Gromicho M, Pinto S, Thompson A, McDermott C, Martin S, Calvo A, Rooney J, van Eijk R, Visser A, Debray T, Kobleva X, Rosenbohm A, Stubendorff B, Hollinger H, Kazoka M, Heverin M, Vajda A, van Rheenen W, Van Vugt J, Dekker A, Middelkoop B, Sommer H, Westeneng H. Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. *Lancet Neurol* 2018; 17:423-433.

Weishaupt J, Weis J, Dorst J, Volk A, Borck G, Sperfeld A, de Carvalho M, Klopstock T, Sendtner M, Otto M, Schuster J, Andersen P, Ludolph A, Strom T, Meitinger T, Freischmidt A, Danzer K, Braak H, Del Tredici K, Jablonka S, Kubisch C, German ALS network MND-NET, Ruf W, Weydt P, Grosskreutz J, Meyer T, Petri S, Grehl T, Müller K, Yilmaz R, Neuwirth C, Weber M, Zeller D, Hübers A, Günther K, Knehr A, Jordan B, Schrank B, Claeys K, Pinto S, Brenner D. Hot-spot KIF5A mutations cause familial ALS. *Brain* 2018

Nordin A, Burkhardt C, Neuwirth C, Holmøy T, Morita M, Tysnes O, Benatar M, Wu J, Lange D, Bisgård C, Asgari N, Tarvainen I, Brännström T, Weber M, Schweikert K, Grehl T, Akimoto C, Wuolikainen A, Alstermark H, Forsberg K, Baumann P, Pinto S, de Carvalho M, Hübers A, Nordin F, Ludolph A, Weishaupt J, Meyer T, Andersen P. Sequence variations in C9orf72 downstream of the hexanucleotide repeat region and its effect on repeat-primed PCR interpretation: a large multinational screening study. *Amyotroph Lateral Scler Frontotemporal Degener* 2016; 18:256-264.

Zhang K, Arcuti S, Brunetti M, Moglia C, Calvo A, Ratti A, Tiloca C, Gellera C, Pensato V, Mazzini L, Capozzo R, Zecca C, Blair I, Stuit R, Muller B, Filosto M, Padovani A, Riva N, Penco S, Lunetta C, Sorarù G, Bertolin C, Blauw H, Curtis C, Hofman A, Estrada K, Rivadeneira F, Uitterlinden A, Dartigues J, Tzourio C, Amouyel P, van der Kooi A, de Visser M, D'Alfonso S, Comi G, Del Bo R, Cereda C, Pansarasa O, Smith B, Shaw C, Weber M, Goris A, Nöthen M, McCann E, Veldink J, Corcia P, Andersen P, Hardiman O, Landers J, Glass J, Brown R, Pers T, Franke L, Van Damme P, Vourc'h P, Silani V, van den Berg L, Al-Chalabi A, Breen G, Lewis C, Pasterkamp R, van Es M, de Bakker P, Visscher P, Wray N, Robberecht W, Weishaupt J, Stubendorff B, Prell T, Ringer T, Witte O, Grosskreutz J, Kiernan M, Pamphlett R, Rowe D, Nicholson G, Kurth I, Hübner C, Ludolph A, Powell J, Logroscino G, Tortelli R, Pupillo E, Beghi E, Chio A, Casale F, Leigh P, Fifita J, Chandran S, Koritnik B, Ravnik-Glavač M, Vrabec K, Rogelj B, Lin K, Ticozzi N, Vajda A, Menelaou A, Medic J, Zidar J, Leonardis L, Polak M, Rojas-García R, Mora J, Pinto S, de Carvalho M, Meininger V, Salachas F, Millecamps S, Grošelj L, Brands W, Schellevis R, Robinson M, de Jong S, Vösa U, van der Spek R, Pulit S, Diekstra F, McLaughlin R, Dekker A, Shatunov A, Yang J, Fogh I, Harschnitz O, van Eijk K, Kenna K, Jones A, Sproviero W, Blokhuis A, Koppers M, Tazelaar G, van Doormaal P, van Rheenen W, Colville S, Cichon S, Maurel C, Andres C, Radivojkov-Blagojevic M, Lichtner P, Meitinger T, Parman Y, Hamzeiy H, Tunca C, Basak A, Bensimon G, Landwehrmeyer B, Rietschel M, Franke A, Lieb W, Tittmann L, Wood N, Dürr A, Saker-Delye S, Payan C, Brice A, McCluskey L, Elman L, Topp S, Malaspina A, Fratta P, Sidle K, Pittman A, Orrell R, Hardy J, Shaw P, Morrison K, Petri S, Abdulla S, Trojanowski J, Van Deerlin V, Lomen-Hoerth C, Wiedau-Pazos M, Staats K, Ophoff R, Meyer T, Sendtner M, Drepper C, Swingler R. Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. *Nat Genet* 2016; 48:1043-8.

Kubisch C, de Carvalho M, Lloyd-Jani A, Konno T, DeJesus-Hernandez M, Angerbauer S, Daoud H, Just W, Tradowsky D, Mouzat K, Landers J, Veldink J, Andersen P, Rademakers R, Van Broeckhoven C, van den Berg L, Rouleau G, Shaw C, Gitler A, Silani V, Nordin A, Calini D, Birve A, Onodera O, Neitzel B, Camu W, Lumbroso S, Leblond C, Van den Broeck M, van Blitterswijk M, Volk A, van Rheenen W, Pinto S, Weber M, Alstermark H, van der Zee J, Ratti A, Chesi A, Keagle P, Talbot K, Proven M, Smith B, Akimoto C. A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. *J Med Genet* 2014; 51:419-24.

## Projekte (0)

Keine Resultate gefunden.

---

Kantonsspital St.Gallen

Rorschacher Strasse 95

CH-9007 St.Gallen

T: +41 71 494 11 11

[support.forschung@kssg.ch](mailto:support.forschung@kssg.ch)