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## Qualifications

## Biography

Robert studied Human Genetics at the University of Nottingham before moving to the Medical Research Council's Human Genetics Unit in Edinburgh to complete his PhD and begin his work using the zebrafish model system. He completed his PhD in 2003 and continued his research at the Victor Chang Cardiac Research Institute in Sydney and the Australian Regenerative Medicine Institute at Monash University. He joined the school as a lecturer and researcher in 2010.

## Research output

### **246th ENMC International Workshop: Protein aggregate myopathies 24–26 May 2019, Hoofddorp, The Netherlands**

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### **Guidelines for the use and interpretation of assays for monitoring autophagy (4th edition)**

Klionsky, D. J., Abdel-Aziz, A. K., Abdelfatah, S., Abdellatif, M., Abdoli, A., Abel, S., Abeliovich, H., Abildgaard, M. H., Abudu, Y. P., Acevedo-Arozena, A., Adamopoulos, I. E., Adeli, K., Adolph, T. E., Adornetto, A., Aflaki, E., Agam, G., Agarwal, A., Aggarwal, B. B., Agnello, M., Agostinis, P. & 230 others, Agrewala, J. N., Agrotis, A., Aguilar, P. V., Ahmad, S. T., Ahmed, Z. M., Ahumada-Castro, U., Aits, S., Aizawa, S., Akkoc, Y., Akoumianaki, T., Akpınar, H. A., Al-Abd, A. M., Al-Akra, L., Al-Gharraibeh, A., Alaoui-Jamali, M. A., Alberti, S., Alcocer-Gómez, E., Alessandri, C., Ali, M., Alim Al-Bari, M. A., Aliwaini, S., Alizadeh, J., Almacellas, E., Almasan, A., Alonso, A., Alonso, G. D., Altan-Bonnet, N., Altieri, D. C., Álvarez, É. M. C., Alves, S., Alves da Costa, C., Alzaharna, M. M., Amadio, M., Amantini, C., Amaral, C., Ambrosio, S., Amer, A. O., Ammanathan, V., An, Z., Andersen, S. U., Andrabi, S. A., Andrade-Silva, M., Andres, A. M., Angelini, S., Ann, D., Anozie, U. C., Ansari, M. Y., Antas, P., Antebi, A., Antón, Z., Anwar, T., Apetoh, L., Apostolova, N., Araki, T., Araki, Y., Arasaki, K., Araújo, W. L., Araya, J., Arden, C., Arévalo, M. A., Arguelles, S., Arias, E., Arikath, J., Arimoto, H., Ariosa, A. R., Armstrong-James, D., Arnauné-Pelloquin, L., Aroca, A., Arroyo, D. S., Arsov, I., Artero, R., Asaro, D. M. L., Aschner, M., Ashrafizadeh, M., Ashur-Fabian, O., Atanasov, A. G., Au, A. K., Auberger, P., Auner, H. W., Aurelian, L., Autelli, R., Avagliano, L., Ávalos, Y., Aveic, S., Avelaira, C. A., Avin-Wittenberg, T., Aydin, Y., Ayton, S., Ayyadevara, S., Azzopardi, M., Baba, M., Backer, J. M., Backues, S. K., Bae, D. H., Bae, O. N., Bae, S. H., Baehrecke, E. H., Baek, A., Baek, S. H., Baek, S. H., Bagetta, G., Bagniewska-Zadworna, A., Bai, H., Bai, J., Bai, X., Bai, Y., Bairagi, N., Baksi, S., Balbi, T., Baldari, C. T., Balduini, W., Ballabio, A., Ballester, M., Balazadeh, S., Balzan, R., Bandopadhyay, R., Banerjee, S., Banerjee, S., Bánrétí, Á., Bao, Y., Baptista, M. S., Baracca, A., Barbati, C., Bargiela, A., Barilà, D., Barlow, P. G., Barmada, S. J., Barreiro, E., Barreto, G. E., Bartek, J., Bartel, B., Bartolome, A., Barve, G. R., Basagoudanavar, S. H., Bassham, D. C., Bast, R. C., Basu, A., Batoko, H., Batten, I., Baulieu, E. E., Baumgarner, B. L., Bayry, J., Beale, R., Beau, I., Beaumatin, F., Bechara, L. R. G., Beck, G. R., Beers, M. F., Begun, J., Behrends, C., Behrens, G. M. N., Bei, R., Bejarano, E., Bel, S., Behl, C., Belaid, A., Belgareh-Touzé, N., Bellarosa, C., Belleudi, F., Belló Pérez, M., Bello-Morales, R., Beltran, J. S. D. O., Beltran, S., Benbrook, D. M., Bendorius, M., Benitez, B. A., Benito-Cuesta, I., Bensalem, J., Berchtold, M. W., Berezowska, S., Bergamaschi, D., Bergami, M., Bergmann, A., Berliocchi, L., Berlioz-Torrent, C., Bernard, A., Berthoux, L., Besirli, C. G., Besteiro, S., Betin, V. M., Beyaert, R., Bezbradica, J. S., Bhaskar, K., Bhatia-Kissova, I., Bhattacharya, R., Bhattacharya, S., Bhattacharyya, S., Bhuiyan, M. S., Bhutia, S. K., Bi, L., Bi, X., Biden, T. J., Bijian, K., Billes, V. A., Binart, N., Bincoletto, C., Birgisdottir, A. B., Bjorkoy, G., Blanco, G., Blas-Garcia, A., Blasiak, J., Blomgran, R., Blomgren, K., Blum, J. S., Boada-Romero, E., Boban, M., Boesze-Battaglia, K., Boeuf, P., Boland, B., Bomont, P., Bonaldo, P., Bonam, S. R., Bonfili, L., Bonifacino, J. S., Boone, B. A., Bootman, M. D., Bordi, M., Borner, C., Bornhauser, B. C., Borthakur, G., Bosch, J., Bose, S., Botana, L. M., Botas, J., Bourke, N. M., Bryson-Richardson, R. J., Furic, L., Harris, J., Pocock, R. & Ruparelía, A. A., 2021, In: *Autophagy*. 17, 1, 381 p.

### **Functional validation of CHMP7 as an ADHD risk gene**

Dark, C., Williams, C., Bellgrove, M. A., Hawi, Z. & Bryson-Richardson, R. J., 6 Nov 2020, In: *Translational Psychiatry*. 10, 1, 8 p., 385.

### **Metformin rescues muscle function in BAG3 myofibrillar myopathy models**

Ruparelía, A. A., McKaige, E. A., Williams, C., Schulze, K. E., Fuchs, M., Oorschot, V., Lacene, E., Meregalli, M., Lee, C., Serrano, R. J., Baxter, E. C., Monro, K., Torrente, Y., Ramm, G., Stojkovic, T., Lavoie, J. N. & Bryson-Richardson, R. J., 19 Oct 2020, (Accepted/In press) In: *Autophagy*. 18 p.

### **BAG3<sup>P215L/KO</sup> Mice as a Model of BAG3<sup>P209L</sup> Myofibrillar Myopathy**

Robertson, R., Conte, T. C., Dicaire, M. J., Rymar, V. V., Sadikot, A. F., Bryson-Richardson, R. J., Lavoie, J. N., O'Ferrall, E., Young, J. C. & Brais, B., Mar 2020, In: American Journal of Pathology. 190, 3, p. 554-562 9 p.

### **KBTD13 is an actin-binding protein that modulates muscle kinetics**

De Winter, J. M., Molenaar, J. P., Yuen, M., Van Der Pijl, R., Shen, S., Conijn, S., Van De Locht, M., Willigenburg, M., Bogaards, S. J. P., Van Kleef, E. S. B., Lassche, S., Persson, M., Rassier, D. E., Sztal, T. E., Ruparelina, A. A., Oorschot, V., Ramm, G., Hall, T. E., Xiong, Z., Johnson, C. N. & 21 others, Li, F., Kiss, B., Lozano-Vidal, N., Boon, R. A., Marabita, M., Nogara, L., Blaauw, B., Rodenburg, R. J., Küsters, B., Doorduyn, J., Beggs, A. H., Granzier, H., Campbell, K., Ma, W., Irving, T., Malfatti, E., Romero, N. B., Bryson-Richardson, R. J., Van Engelen, B. G. M., Voermans, N. C. & Ottenheijm, C. A. C., 3 Feb 2020, In: Journal of Clinical Investigation. 130, 2, p. 754-767 14 p.

### **A transgenic zebrafish model of hepatocyte function in human Z $\alpha$ 1-antitrypsin deficiency**

Yip, E., Giousoh, A., Fung, C., Wilding, B., Prakash, M. D., Williams, C., Verkade, H., Bryson-Richardson, R. J. & Bird, P. I., 12 Jun 2019, In: Biological Chemistry. 400, 12, p. 1603-1616 14 p.

### **Linking life-history theory and metabolic theory explains the offspring size-temperature relationship**

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### **Metformin rescues muscle function in BAG3 myofibrillar myopathy models**

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### **Recent advances in understanding congenital myopathies**

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### **Advances in the Understanding of Skeletal Myopathies from Zebrafish Models**

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### **The role of ADHD associated genes in neurodevelopment**

Dark, C., Homman-Ludiye, J. & Bryson-Richardson, R. J., 15 Jun 2018, In: Developmental Biology. 438, 2, p. 69-83 15 p.

### **Testing of therapies in a novel nebulin nemaline myopathy model demonstrate a lack of efficacy**

Sztal, T. E., McKaige, E. A., Williams, C., Oorschot, V., Ramm, G. & Bryson-Richardson, R. J., 30 May 2018, In: Acta Neuropathologica Communications. 6, 10 p., 40.

### **Does the cost of development scale allometrically with offspring size?**

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### **Genetic compensation triggered by actin mutation prevents the muscle damage caused by loss of actin protein**

Sztal, T. E., McKaige, E. A., Williams, C., Ruparelina, A. A. & Bryson-Richardson, R. J., 1 Feb 2018, In: PLoS Genetics. 14, 2, 15 p., e1007212.

### **L-tyrosine supplementation does not ameliorate skeletal muscle dysfunction in zebrafish and mouse models of dominant skeletal muscle $\alpha$ -actin nemaline myopathy**

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### **Genome-wide identification of conserved intronic non-coding sequences using a Bayesian segmentation approach**

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### **Analysis of RNA expression in adult Zebrafish skeletal muscle**

Sztal, T. E., Currie, P. D. & Bryson-Richardson, R. J., 2017, *Skeletal Muscle Development*. Humana Press, Vol. 1668. p. 27-35 9 p. (Methods in Molecular Biology; vol. 1668).

### **Production of zebrafish cardiospheres and cardiac progenitor cells in vitro and three-dimensional culture of adult zebrafish cardiac tissue in scaffolds**

Zeng, W. R., Beh, S.-J., Bryson-Richardson, R. J. & Doran, P. M., 2017, In: *Biotechnology and Bioengineering*. 114, 9, p. 2142–2148 7 p.

### **Variants in the oxidoreductase PYROXD1 cause early-onset myopathy with internalized nuclei and myofibrillar disorganization**

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### **Using touch-evoked response and locomotion assays to assess muscle performance and function in zebrafish**

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### **Filamin C is a highly dynamic protein associated with fast repair of myofibrillar microdamage**

Leber, Y., Ruparelia, A. A., Kirfel, G., van der Ven, P. F. M., Hoffmann, B., Merkel, R., Bryson-Richardson, R. J. & Furst, D. O., 1 Jul 2016, In: *Human Molecular Genetics*. 25, 13, p. 2776-2788 13 p.

### **FLNC myofibrillar myopathy results from impaired autophagy and protein insufficiency**

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### **Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition)**

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### **Bone morphogenetic protein/retinoic acid inducible neural-specific protein (brinp) expression during danio rerio development**

Gioush, A., Rodrigues Vaz, R., Bryson-Richardson, R. J., Whisstock, J. C., Verkade, H. M. & Bird, P. I., 2015, In: *Gene Expression Patterns*. 18, 1-2, p. 37 - 43 7 p.

### **Comparison of different numerical treatments for x-ray phase tomography of soft tissue from differential phase projections**

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### **Zebrafish models for nemaline myopathy reveal a spectrum of nemaline bodies contributing to reduced muscle function**

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### **Zebrafish models of BAG3 myofibrillar myopathy suggest a toxic gain of function leading to BAG3 insufficiency**

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### **In vivo wall shear measurements within the developing zebrafish heart**

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### **Student acceptance and application of peer assessment in a final year genetics undergraduate oral presentation**

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### **Atlas of Zebrafish Development**

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### **Cardiac-phase filtering in intracardiac particle image velocimetry**

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