

Dear Sir/Madam,

We are writing to you to ask for your assistance in addressing the misconception that males with Rett syndrome do not survive beyond infancy. Not only is the distressing to read for parents of male children with Rett syndrome, it also is detrimental to them receiving appropriate and timely healthcare because uninformed professionals (who will google the condition) have the belief that active treatment is not warranted, they are doing to die anyway. This is the worst case scenario, as you can imagine I am sure but it happens.

It is also detrimental to them accessing appropriate services and therapies e.g. physiotherapy, speech and language therapy and occupational therapy but there are others that help improve their quality of life.

Providers of funding for health and social care, again who are not familiar with the disorder and how it presents in males, are also reluctant to put respite and care packages in place and to forward plan because again they are under the misconception that this will not be necessary.

I am grateful to my colleague and fellow parent (of a male with Rett syndrome) who has assisted in putting together some resources including research papers that all corroborate what I am saying.

I understand that the mum concerned, Mary Engel, contacted you a couple of years ago to raise this with you and in fact, did an interview with yourselves. Kate Fernindansen also contributed. Yet sadly, this remains an issue with the publications and stories you share.

Kate adds

'We have numerous parents and caretakers of males, willing to confirm that their son's have a Rett diagnosis and do exist beyond infancy. Many of us can also provide concrete examples of how mis-information has directly impacted or delayed our son's diagnosis and care and that has to change.

I am willing to discuss this population, as evidenced by the interview I did for Rett Syndrome News 2 years ago along with the mother of another boy who has Rett, Ms. Mary Engel. You can Google her name along with "Rett syndrome" in order to see evidence that her adorable son, Henry, does exist and is no longer an infant.

I am eager to work toward a new era in which information provided to the public about our son's is updated and accurate, so that boys affected by Rett syndrome in the future will receive help without questions or delay. This begins by rectifying public misperceptions via a collaboration of efforts. We hope that Rett Syndrome News will wholeheartedly join us in our mission. Thank you.'

I hope this helps Rett Syndrome News understand not only that males with Rett syndrome can live beyond infancy but also why perpetuating this outdated notion is so detrimental to families, care givers and males living with the disorder.

Yours sincerely,

A handwritten signature in black ink, appearing to read 'Jen' followed by a dash.

Mrs R Jenner
President
Rett Syndrome Europe

A handwritten signature in black ink, appearing to read 'Robert Adamek'.

Mr Robert Adamek
CEO
Rett UK

Resources on Males with Rett Syndrome

Organisations that have information on males with Rett on their websites are:

[The International Rett Syndrome Foundation \(Rettsyndrome.org\)](http://Rettsyndrome.org) – Dad of a male child with Rett syndrome joins the board

[Rett UK](#) – FAQs – why does Rett syndrome mainly occur in females?

[RSRT](#) – Genetics Primer - Can Males Have Rett Syndrome?

[Rett Syndrome Europe](#) – Can Boys Have Rett Syndrome?

There are over 20 published research papers on males with Rett syndrome and all are available on reference sites such as Pubmed, Google Scholar.

Here are 2 examples...

The array of clinical phenotypes of males with mutations in Methyl-CpG binding protein 2

doi: 10.1002/ajmg.b.32707

Neul JL, Benke TA, Marsh ED, Skinner SA, Merritt J, Lieberman DN, Standridge S, Feyma T, Heydemann P, Peters S, Ryther R, Jones M, Suter B, Kaufmann WE, Glaze DG, Percy AK. The array of clinical phenotypes of males with mutations in Methyl-CpG binding protein 2. *Am J Med Genet B Neuropsychiatr Genet.* 2019 Jan;180(1):55-67. doi: 10.1002/ajmg.b.32707. Epub 2018 Dec 7. PMID: 30536762; PMCID: PMC6488031.

Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements

doi: 10.1111/cge.13473.

Schönewolf-Greulich B, Bisgaard AM, Dunø M, Jespersgaard C, Rokkjaer M, Hansen LK, Tsoutsou E, Sofokleous C, Topcu M, Kaur S, Van Bergen NJ, Brøndum-Nielsen K, Larsen MJ, Sørensen KP, Christodoulou J, Fagerberg CR, Tümer Z. Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. *Clin Genet.* 2019 Mar;95(3):403-408. doi: 10.1111/cge.13473. Epub 2018 Dec 7. PMID: 30417326.

The International Rett Syndrome Foundation's YouTube Channel has a webinar in which Texas Children's Rett research physician, Dr. Bernhard Suter, explains the different ways males can have Rett syndrome called "[Let's Hear it for the Boys! Males with MECP2 Mutations.](#)"

Here are some of the Rett syndrome research physicians who are able to confirm that males diagnosed with Rett syndrome do exist beyond the age of infancy and some, even into adulthood.

They are; Dr. Alan K. Percy of the University of Alabama, US, Dr. Jeffrey Neul of Vanderbilt University, US, Dr. Timothy Benke, of the University of Colorado, US, Dr. Bernhard Suter & Dr. Davut Pehlivan, of Texas Children's Hospital, Dr. Eric Marsh, Children's Hospital of Philadelphia, Dr. Steven Skinner of Greenwood Genetics Center, US



Here are some Rett non profit organisations that are aware and can also confirm that boys and men diagnosed with Rett syndrome do exist beyond infancy.

Ms. Melissa Kennedy, CEO of the International Rett Syndrome Foundation

Ms. Monica Coenraads, CEO of Rett Syndrome Research Trust

Ms. Rachael Stevenson, CEO Reverse Rett UK