

#### No.\*, 29th Month, 2019

#### Welcome to new staff

Your Library is a partner in the health care team, and provides the link between information and its application in patient care, education, and research

Our goal is to help you to access the information you need whenever you need it and from wherever you are.

Finding information, no matter how obscure, is what we do.

#### Facilities

- 24/7 swipe access
- Print, scan, photocopying, and other <u>document services</u>
- 14 desktop computers to use in the Library, or bring your own device and connect to Wi-Fi
- Study areas
- Book Swap: fiction and non-fiction titles, free to use, no membership required. If you take a book, please leave one of your own in its place.

#### Resources

- Online and in print journals and books
- Databases, Australian and International Standards, drug information
- <u>Remote access</u>
- EndNote: borrow a disk and install it at work or home
- <u>Fact Sheets</u>: our collection of cheat sheets on database searching and using Library services

#### Services

- <u>Training</u>, including *Introduction to the Library*, *EndNote*, *Ovid Medline*, *Nursing & Allied Health resources*, and *PubMed*
- <u>Document Delivery</u>: request journal articles, book chapters, or books (requires registration)
- <u>Literature search</u> assistance, including systematic reviews
- <u>Ask a Librarian</u>: ask us a question and get a response within 1 business day







Share this Library Update with your new colleagues and encourage them to subscribe

# PubMed: new interface, and how to show links to full text content

The <u>new PubMed</u> will become the default in 2020, and will ultimately replace the <u>current version</u>

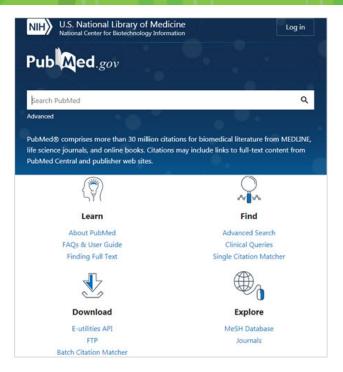
It features a responsive layout offering better support for mobiles and tablets, and allows you to find highly relevant articles with the default *Best Match* sort.

The National Library of Medicine will continue adding features and improving the user experience during the testing phase.

At this stage, searching via Medical Subject Headings (MeSH) is not as straightforward as in the current interface, so it is not well suited to complex searches.

Customise PubMed to get direct links to Library-subscribed full text content

One new feature that the Library is already taking



advantage of is *LinkOut*. It is a vastly improved method of linking from PubMed abstracts to Library-subscribed full text content. You can use either our <u>customised PubMed link</u> on our <u>databases list</u> or configure your *MyNCBI* account to see full text links.

When you don't find the full text article you can click straight through to a pre-filled <u>Document Delivery</u> request form.

See our Fact Sheet no.14 PubMed for details, or contact us for help.

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Presentation and Treatment of Histoplasmosis in Pediatric Oncology Patients: Case Series and Review of the Literature. Histopl Constitution Mark Science & Author Information Save Items	Title: Presentation and Treatment of Histoplasmosis in Pediatric Oncology Patient: Source: Journal of Pediatric Hematology/Oncology [1077-4114] Hess, Jennifer yr:20
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Follow the link to get full text, or use <b>Document Delivery</b> to order your article if no online or print sources are found.	Year: 2017 Volume: 39 Issue: 2 Start Page: 137 Go Can't find it?     Order an article via Document Delivery Go

#### **BMJ Best Practice**

BMJ Best Practice brings a step-by-step approach to decision support. It is structured around the patient consultation, covering diagnosis, prognosis, treatment, and prevention.

- <u>Atypical pneumonia</u>
- <u>Delayed puberty</u>
- Developmental dysplasia of the hip
- <u>Enuresis</u>
- <u>Infantile spasms</u>
- <u>Premature newborn care</u>

- <u>Pyloric stenosis</u>
- Sepsis in children
- <u>Severe combined immunodeficiency</u>
- Type 1 diabetes
- Viral gastroenteritis in children

#### Product trial reminder: DynaMed

# Make the most of our trial of <u>DynaMed</u> before 31<sup>st</sup> March 2020

Create a personal account to enable seamless access outside the RCH network and via the app on your mobile device, as well as allowing CME tracking across devices and platforms. You can also follow topics, and be alerted to practice changing and other updates.

# Articles relating to <u>Novel Coronavirus (2019-nCoV</u>) are free, regardless of subscription status.

#### Is DynaMed a true alternative to UpToDate?

The Health Sciences Library at Penn State College of Medicine recently <u>compared the two</u>, and found they are aligned in all areas.

#### Provide us with feedback about your experience

We urge you to provide us with genuine feedback about this product. Does it meet your expectations? Is it intuitive to use? You can let us know <u>here</u>.

### Health awareness calendar

- Feb Ovarian Cancer Awareness Month
- Feb <u>Show Your Heart: FebUnplugged</u>
- Feb 2–8 Feeding Tube Awareness Week
- Feb 4 <u>World Cancer Day</u>
- Feb 11 <u>Safer Internet Day</u>
- Feb 18 International Asperger's Day
- Feb 20 <u>World Day of Social Justice</u>
- Feb 22 <u>World Encephalitis Day</u>
- Mar 3 <u>World Hearing Day</u>
- Mar 1-7 <u>Hearing Awareness Week</u>

### Library Training: Session times for February/March 2020

If you can't make it to the scheduled sessions, <u>contact us</u> to arrange individual or group sessions at a time that suits you. **Charges apply for some Library training**. <u>See the website for more details</u>.

Session	February	March
Nursing & Allied Health	-	Wednesday 18 March, 2:00 pm
Cochrane Library	-	Monday 2 Mar, 10:00 am
EndNote Tutorial 1	Wednesday 12 February, 2:30 pm	Tuesday 10 March, 10:00 am
EndNote Tutorial 2	Wednesday 26 February, 2:30 pm	Tuesday 17 March, 10:00 am
EndNote Demonstration	-	Wednesday 18 Mar, 8:30 am
Finding the Evidence	-	Monday 2 March, 9:00 am
Introduction to the Library	Thursday 13 February, 9:15 am	Tuesday 24 March, 12:00 pm
Ovid Medline	-	Tuesday 10 March, 12:00 pm
PubMed	Monday 10 February, 8:30 am	Monday 16 March, 10:30 am

### What's new from AIHW

#### Australia's children

Childhood is an important time for healthy development, learning, and establishing the foundations for future wellbeing. Most Australian children are healthy, safe and doing well. However, childhood is also a time of vulnerability and a child's outcomes can vary depending on where they live and their family's circumstances. This report brings together a range of data on children's wellbeing and their experiences at home, school and in the community. <u>Read now</u>.

#### Alcohol, tobacco & other drugs in Australia

The consumption of alcohol, tobacco and other drugs is a major cause of preventable disease and illness in Australia. This report consolidates the most recently available information on alcohol, tobacco and other drug use in Australia, and includes key trends in the availability, consumption, harms and treatment for vulnerable populations. Further, information on a range of health, social and economic impacts of alcohol, tobacco and other drug use are highlighted. <u>Read now</u>.

#### **Health and Science News**

- WHO declares the new coronavirus outbreak a Public Health Emergency of International Concern: The WHO Director-General made the declaration following the advice of the Emergency Committee. <u>Read more</u> [source: who.int]
- Australian lab first outside of China to re-create coronavirus, helping vaccine push: Described as a "game changer", this means scientists are able to test potential vaccines and to develop a test to identify people who might be infected. <u>Read more</u> [source: abc.net.au]
- University of Queensland asked to rapidly develop coronavirus vaccine: The Coalition for Epidemic Preparedness Innovations (CEPI) has requested the university use its recently developed rapid response technology to develop a new vaccine. <u>Read more</u> [source: hospitalhealth.com.au]
- Wiley provides free access to coronavirus research to aid relief efforts: These important pieces of literature will remain free until April 2020 and the window of time will be extended as needed. As new articles are published, they will be made free to access. <u>Read more</u> [source: eurekalert.org]
- Predatory-journal papers have little scientific impact: A study showed that around 60% of the papers hadn't attracted any citations at all, and 38% were cited up to 10 times. Less than 3% of the papers attracted more than 10 citations, and none got more than 32 citations. <u>Read more</u> [source: nature.com]
- Nearly all U.S. kids eating added sugars before age two: The study team found that 98.3% of toddlers (ages 12 months to 23 months) and 60.6% of infants (age up to 11 months) consumed added sugars, mainly from yogurt, baby food, snacks and sweets, bakery products and fruit drinks. <u>Read</u> <u>more</u> [source: reuters.com]
- Trial suggests babies in intensive care can be better protected from parental bacteria: Johns Hopkins Medicine researchers turned to a simple regimen for mothers and fathers to follow while their child is in intensive care. <u>Read more [source:</u> eurekalert.org]
- Old whooping cough vaccine could protect against food allergies: A review of child food allergy cases from the late 1990s has revealed that children who received one or more doses of an older whole-cell whooping cough vaccine were 23% less likely to be diagnosed with a food allergy than those who had the updated acellular version. <u>Read</u> <u>more</u> [source: hospitalhealth.com.au]

- What's next for nursing? How a changing paradigm is impacting the profession: An independent survey of nearly 2,000 consumers, nurses, doctors, and healthcare executives in the U.S. provides insights into the top trends that will shape priorities over the next few years. <u>Read</u> <u>more</u> [source: electronichealthreporter.com]
- Giving premature babies 100% oxygen may help them breathe on their own faster: This careful balancing act of stimulating babies to breathe with excess oxygen, and then dialling it back once it's no longer needed could save lives in the future. <u>Read</u> more [source: scimex.org]
- ⇒ FDA and NIH let clinical trial sponsors keep results secret and break the law: For 20 years, the U.S. government has urged companies, universities, and other institutions that conduct clinical trials to record their results in a federal database, so doctors and patients can see whether new treatments are safe and effective. Few trial sponsors have consistently done so. <u>Read more</u> [source: sciencemag.org]
- Prescription drug improves symptoms of autism by targeting brain's chemical messengers: A prescription drug for oedema iimproves some of the symptoms in young children with autism spectrum disorders and has no significant side effects, according to a new study. <u>Read more</u> [source: eurekalert.org]
- Chinese scientist who produced genetically altered babies sentenced to 3 years in jail: The court ruled that the three defendants had deliberately violated national regulations on biomedical research and medical ethics, and rashly applied gene-editing technology to human reproductive medicine. <u>Read more</u> [source: sciencemag.org]
- App helps parents of premature babies stay in touch: Doctors and nurses can send photos and video updates to parents when they can't be with their baby in hospital. <u>Read more</u> [source: bbc.com]
- The best science images of the year: 2019 in pictures: A black hole's perilous edge, antipoaching warriors, whisky webs and more. <u>Read</u> <u>more</u> [source: nature.com]
- ➡ Epic results for preemie baby survival rates: The rate of survival of very preterm babies in Canada increased 25% after the national Evidence-based Practice for Improving Quality (EPIQ) program was introduced in neonatal intensive care units (NICUs) across the country. <u>Read more [source: scimex.org]</u>

#### EndNote Tip: Upgrade to the latest version on PC and Mac

Apple has dropped all 32-bit application support, with implications for both PC and Mac users of EndNote

The easiest way to avoid incompatibilities is to ensure you are using the latest version of EndNote on all your devices. That's version X9.3.1 for Windows and X9.3.2 for Mac. Open EndNote and go to Help/About to find your current version.

# You must uninstall prior versions of EndNote before installing X9.

**If your EndNote version is prior to X9** you can borrow an install disk from the Library, or <u>contact us</u> if you have no disk drive. **If you are already on X9** you can <u>upgrade here</u>.

Version X9.3 onwards for Mac is compatible with macOS Catalina\*. Version X9.3 onwards for Windows is cross-compatible with X9.3.2 Mac.

#### After upgrading

When you first open your EndNote library that was created with an older version, you will receive the following message: "This library was created using an earlier version of EndNote and needs to be converted for use with this version of EndNote. A new, converted library will be created that can only be used with X9.3 and later, but the original library will not be changed. To continue using the library in X9.2 and earlier, use the original library or use EndNote sync. Continue with the library conversion"

Your old library will be converted and be renamed to *My EndNote library-Converted.enl*.

If you try to open this converted library or any library created on X9.3 onwards on an earlier version of EndNote, it will open a blank library.



\* It is expected that macOS 10.15 Catalina will not run EndNote X8 and earlier. If you wish to continue using EndNote X8, do not upgrade to Catalina.

#### **Top 50 Most-Cited Campus Publications - updated**

<u>This list</u> is updated every three to four months; significant changes are unlikely to occur more frequently.

We compile the list using *Scopus*, an abstract and citation database of peer-reviewed literature.

We search the Affiliation field for *Royal Children's Hospital, Murdoch Childrens Research Institute,* and

Department of Paediatrics, University of Melbourne. Results are then sorted in descending order of times cited.

Each item is manually edited for formatting consistency, The list is then exported and converted to a usable format

#### CAMPUS PUBLICATIONS

Article titles are linked to full text where available, or to information about ordering documents. An OpenAthens account is required for remote access.

Akesson, L., Fennell, A., Bournazos, A., Krzesinski, E., Tan, K., Springer, A., Rose, K.,
 Goranitis, I., Lee, C., Christodoulou, J., Lunke, S., Stark, Z., Hunter, M. and Cooper, S.
 [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Rapid mRNA splicing analysis confirming pathogenicity of a novel homozygous ASNS variant in a newborn',
 <u>Twin Research and Human Genetics 2019; vol.22(5), pp.371</u>

Akesson, L., Oliver, M., Cole, T., Simm, P., Francis, D. and White, S. [Department of Endocrinology, Department of Gastroenterology, Department of Immunology, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) caused by a homozygous intragenic AIRE deletion detected by chromosomal microarray', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.372</u>

- Allen, W.M.K. and Allen, K.J. [Population Health Research Theme, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Should Australia tax sugar-sweetened beverages?', *Journal of Paediatrics and Child Health 2019; [Epub ahead of print]*
- Amor, D., Stephenson, S., Mustapha, M., Mensah, M., Ockeloen, C., Lee, W., Tankard, R., Phelan, D., Shinawi, M., De Brouwer, A., Pfundt, R., Dowling, C., Toler, T., Sutton, R., Agolini, E., Rinelli, M., Capolino, R., Martinelli, D., Zampino, G., Dumic, M., Reardon, W., Shaw-Smith, C., Leventer, R., Delatycki, M., Kleefstra, T., Mundlos, S., Mortier, G., Bahlo, M., Allen, N. and Lockhart, P. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Pathogenic variants in GPC4 cause Keipert syndrome', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.337</u>
- Andersen, T., Greaves, R., McWhinney, A., Massie, J., Matthews, S., Shepherd, S., Webster,
   D., Wiley, V. and Graham, P. [Victorian Clinical Genetics Services, Murdoch Childrens Research Institute; Respiratory and Sleep Medicine, Royal Children's Hospital,
   Flemington Road, Parkville, Vic., Australia.], 'Establishment of an Asia-Pacific dried blood spot external quality assurance program', <u>Twin Research and Human Genetics 2019;</u> vol.22(5), pp.407-408
- Aslam, H., Holloway-Kew, K.L., Mohebbi, M., Jacka, F.N. and Pasco, J.A. [Centre for Adolescent Health, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Association between dairy intake and fracture in an Australian-based cohort of women: a prospective study', <u>BMJ Open 2019; vol.9(11), pp.e031594</u>
- Attard, C. and Ignjatovic, V. [Haematology Research Laboratory, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Primary thromboprophylaxis in children with cancer: a road less travelled', <u>Thrombosis</u> <u>and Haemostasis 2019; vol.119(12), pp.1894-1896 (full text via Document Delivery)</u>
- Babl, F.E., Cheek, J., Dalziel, K., Bressan, S. and Dalziel, S.R. [Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'In reply', <u>Annals of Emergency</u> <u>Medicine 2019; vol.74(6), pp.821-823</u>
- Bahlo, M., Bennett, M., Rafehi, H., Wallis, M., Christodoulou, J., Hildebrand, M., Brown, N., Berkovic, S., Mullen, S., White, S., Delatycki, M. and Lockhart, P. [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Detecting repeat expansions with sequencing data: implications for novel discoveries and rapid genetic diagnosis', <u>Twin Research and Human Genetics 2019</u>; vol.22(5), pp.345-346
- Bannister, E., Sozzi, V., Mason, H., Locarnini, S., Hardikar, W. and Revill, P.A. [Department of Gastroenterology and Clinical Nutrition, Royal Children's Hospital; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Analysis of the in vitro replication phenotype of African hepatitis B virus (HBV) genotypes and subgenotypes present in Australia identifies marked differences in DNA and protein expression', *Virology 2020; vol.540, pp.97-103*
- Berecki-Gisolf, J., Rowland, B., Reavley, N., Minuzzo, B. and Toumbourou, J. [Centre for Adolescent Health, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Evaluation of community coalition training effects on youth hospital-admitted injury incidence in Victoria, Australia: 2001-2017', <u>Injury Prevention 2019; [Epub ahead of print]</u>
- Best, S., Long, J., Stark, Z., Gaff, C., Braithwaite, J. and Taylor, N. [Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Clinical leadership and genomics - what works?', *Twin Research and Human Genetics 2019; vol.22(5), pp.350*
- Boniface, K., Byars, S.G., Cowley, D., Kirkwood, C.D. and Bines, J.E. [Enteric Diseases Group, Murdoch Childrens Research Institute; Department of Gastroenterology and Clinical Nutrition, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Human neonatal rotavirus vaccine (RV3-BB) produces vaccine take irrespective of histo-blood group antigen status', *Journal of Infectious Diseases 2019; Nov 25 [Epub ahead of print]*

- Borschmann, R., Molyneaux, E., Spry, E., Moran, P., Howard, L.M., Macdonald, J.A., Brown, S.J., Moreno-Betancur, M., Olsson, C.A. and Patton, G.C. [Centre for Adolescent Health, Clinical Epidemiology and Biostatistics Unit; Healthy Mothers Healthy Families, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Pre-conception self-harm, maternal mental health and mother-infant bonding problems: a 20-year prospective cohort study', <u>Psychological Medicine 2019; vol.49(16), pp.2727-2735</u>
- **Bowman-Smart, H., Gyngell, C., Morgan, A. and Savulescu, J.** [Biomedical Ethics Research Group, Speech and Language Group, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'The moral case for sign language education', <u>Monash Bioethics Review 2019; vol.37(3-4), pp.94-110</u>
- **Bowman-Smart, H., Savulescu, J., Delatycki, M., Mand, C. and Gyngell, C.** [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'The Ethics of NIPT: women's experiences and attitudes on expanding the reach', *Twin Research and Human Genetics 2019; vol.22(5), pp.347*
- Boys, A., Smith, M., Wei, V., Monaco, A., Yiu, E., Krishnamurthy, P., Marum, J., Petrovic, V., Curnow, L. and Delatycki, M. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'The elusive Spinal Muscular Atrophy (SMA) diagnosis: homozygous sequence variants', *Twin Research and Human Genetics 2019; vol.22(5), pp.379*
- Brett, G., Martyn, M., De Silva, M., Boggs, K., Baxendale, A., Borrie, S., King-Smith, S., Ayres, S., Gallacher, L., Lynch, F., Pinner, J., Sandaradura, S., Wilson, M., Barnett, C., Patel, C., Vasudevan, A., Krzesinski, E., Lunke, S. and Stark, Z. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Parent experiences with ultra-rapid genomic sequencing in pediatric acute care', <u>Twin Research and Human Genetics 2019</u>; vol.22(5), pp.349
- Breuer, O., Schultz, A., Garratt, L.W., Turkovic, L., Rosenow, T., Murray, C.P., Karpievitch, Y.V., Akesson, L., Dalton, S., Sly, P.D., Ranganathan, S., Stick, S.M. and Caudri, D. [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Aspergillus infections and progression of structural lung disease in children with cystic fibrosis', <u>American Journal</u> of Respiratory and Critical Care Medicine 2019; [Epub ahead of print]
- Cai, S., Thompson, D.K., Anderson, P.J. and Yang, J.Y. [Victorian Infant Brain Study, Developmental Imaging, Neuroscience Research, Murdoch Childrens Research Institute; Department of Neurosurgery, Royal Children's Hospital; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Short- and longterm neurodevelopmental outcomes of very preterm infants with neonatal sepsis: a systematic review and meta-analysis', *Children 2019; vol.6(12), article no. 131*
- **Choy, C., Sporton, A., Head-Gray, J., Senanayake, S., Raj, S. and Savarirayan, R.** [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Life at the coalface: a clinical trial coordinator's perspective', *Twin Research and Human Genetics 2019; vol.22(5), pp.414*
- Christodoulou, J. [Brain and Mitochondrial Research Group, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Therapeutic advances in the management of mitochondrial disorders', <u>Twin Research</u> <u>and Human Genetics 2019; vol.22(5), pp.334</u>
- **Cloney, T., White, S., Gallacher, L., Christodoulou, J., Pais, L., Elliott, J. and Tan, T.** [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'The diagnostic trajectory of families undiagnosed after singleton exome sequencing', *Twin Research and Human Genetics 2019; vol.22(5), pp.340*
- **Collie, J.T.B., Greaves, R.F., Jones, O.A.H., Eastwood, G. and Bellomo, R.** [Victorian Clinical Genetic Services, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Vitamin C measurement in critical illness: challenges, methodologies and quality improvements', <u>*Clinical Chemistry and Laboratory Medicine 2019; Dec 12 [Epub ahead of print]*</u>

- Compton, A., Frazier, A., Thirukeswaran, S., Lake, N., Amarasekera, S., Rius, R., Cummings, B., Pais, L., Tucker, E., Mountford, H., Lim, S., White, S., Simons, C., Christodoulou, J. and Thorburn, D. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Outside the exome: the role of CNVS, SVS, deep-intronic and 'silent' mutations in mitochondrial disease', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.344-345</u>
- Cowie, S., Lake, N., Love, C., Pantaleo, S., Chong, B., Compton, A., Thorburn, D., Christodoulou, J. and Lunke, S. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Whole mitochondrial genome sequencing at VCGS', *Twin Research and Human Genetics 2019; vol.22(5), pp.368*
- Cusack, M., Hickerton, C., Dunlop, K., Nisselle, A., McClaren, B., Terrill, B., Robinson, F., Pirotta, M. and Metcalfe, S. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'General practitioners and genomics: their views on practice and education in Australia', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.342-343</u>
- Dadds, M.R., Sicouri, G., Piotrowska, P.J., Collins, D.A.J., Hawes, D.J., Moul, C., Lenroot, R.K., Frick, P.J., Anderson, V., Kimonis, E.R. and Tully, L.A. [Child Neuropsychology, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Keeping parents involved: predicting attrition in a self-directed, online program for childhood conduct problems', *Journal of Clinical Child and Adolescent Psychology 2019; vol.48(6), pp.881-893*
- Damiano, J.A., Deng, L., Li, W.H., Burgess, R., Schneider, A.L., Crawford, N.W., Buttery, J., Gold, M., Richmond, P., Macartney, K.K., Hildebrand, M.S., Scheffer, I.E., Wood, N. and Berkovic, S.F. [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'SCN1A variants in vaccine-related febrile seizures: a prospective study', <u>Annals of Neurology 2019; [Epub ahead of print]</u>
- D'Aprano, A., Riess, S., Cooper, M., Meehan, E., Mountford, N. and Gibb, S. [Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Important components of a programme for children with medical complexity: an Australian perspective', <u>Child:</u> <u>Care, Health and Development 2020; vol.46(1), pp.90-103</u>
- De Silva, M., Brett, G., Ayres, S., Baxendale, A., Borrie, S., Boggs, K., Springer, A., Rose, K., Cunningham, F., Curd, H., Davis, T., Fowles, L., Hunt, L., Lynch, F., Wilson, M., Stark, Z. and Gallacher, L. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Genetic counselors in the NICU and PICU: experiences from the Australian Acute Care Genomics project', *Twin Research and Human Genetics 2019; vol.22(5), pp.350*
- De Valles-Ibanez, G., Poke, G., King, C., Muir, A., Germano, M., De Souza, C.M., Fung, J., Chung, B., Fung, C.W., Mignot, C., Ilea, A., Keren, B., Davis, S., Stanley, T., Moharir, M., Kannu, P., Shao, Z., Malerba, N., Merla, G., Mefford, H., Scheffer, I. and Sadleir, L. [Royal Children's Hospital; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'The epileptology of GNB5 encephalopathy', *Twin Research and Human Genetics 2019; vol.22(5), pp.405*
- **Delatycki, M., Kirk, E. and Laing, N.** [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Mackenzie's mission', *Twin Research and Human Genetics 2019; vol.22(5), pp.330*
- Delatycki, M.B., Alkuraya, F., Archibald, A., Castellani, C., Cornel, M., Grody, W.W., Henneman, L., Ioannides, A.S., Kirk, E., Laing, N., Lucassen, A., Massie, J., Schuurmans, J., Thong, M.K., van Langen, I. and Zlotogora, J. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'International perspectives on the implementation of reproductive carrier screening', <u>Prenatal Diagnosis 2019; [Epub ahead of print]</u>
- Dionne, A., Dahdah, N., Singh-Grewal, D., Burgner, D.P., Newburger, J.W. and de Ferranti, S.D. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Anti-thrombosis management of patients with Kawasaki disease: results from an international survey', <u>International Journal of</u> <u>Cardiology 2019; [Epub ahead of print]</u>

- Downie, L., Halliday, J., Lunke, S., Lynch, E., Martyn, M., Gaff, C., Hunter, M., Lewis, S., Jarmolowicz, A. and Amor, D. [Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Baby beyond hearing, using genomics as a newborn screening tool', <u>Twin Research and Human Genetics 2019; vol.22(5)</u>, <u>pp.351</u>
- Ede, J., Jeffs, E., Vollam, S. and Watkinson, P. [Emergency Department, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'A qualitative exploration of escalation of care in the acute ward setting', <u>Nursing in Critical Care 2019; Dec 12 [Epub</u> <u>ahead of print]</u>
- Elliott, J., Chong, B., White, S., Tan, T. and Lunke, S. [Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Implementation of exome sequencing in a clinical diagnostic laboratory service', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.339</u>
- Elliott, J., Hanna, C., Ayers, K., Robevska, G., Van Den Bergen, J., Phillimore, H., Flanagan, D., Pantaleo, S.J., Grover, S., O'Connell, M. and Sinclair, A. [Murdoch Childrens Research Institute; Royal Children's Hospital; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Use of a diagnostic DSD gene panel and implications for clinical care', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.408</u>
- **Evans, M., Fitzell, K. and Normoyle, M.** [Department of Metabolic Medicine, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'The use of tetrahydrobiopterin in PKU: the Victorian experience', *Twin Research and Human Genetics 2019; vol.22(5), pp.353-354*
- Fearn, N.D. and Mackay, M.T. [Department of Neurology, Royal Children's Hospital; Clinical Sciences Research Theme, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Focal cerebral arteriopathy and childhood stroke', <u>Current Opinion in Neurology 2019; Dec 6</u> [Epub ahead of print]
- Ferguson, G., Simm, P., O'Connell, M. and Pang, K.C. [Royal Children's Hospital; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Gender dysphoria: puberty blockers and loss of bone mineral density', <u>BMJ (Clinical research ed.)</u> 2019; vol.367, pp.16471
- Fitzell, K. and Evans, M. [Department of Nutrition and Food Services, Department of Metabolic Medicine, Royal Children's Hospital Flemington Road, Parkville, Vic., Australia.], 'Continuous glucose monitoring in hepatic glycogen storage disorders: a systematic review', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.353</u>
- Flowers, N., Love, C., Giouzeppos, O., Shi, G., Hunt, C., Scarff, K., Archibald, A. and Pertile,
   M. [Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.],
   'Improved positive predictive value for monosomy X using a cell-free DNA paired-end
   sequencing assay', *Twin Research and Human Genetics 2019; vol.22(5), pp.366-367*
- Forbes Shepherd, R., Keogh, L., Werner-Lin, A., Delatycki, M. and Forrest, L. [Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'How do adolescents and young adults experience genetic testing for Li-Fraumeni syndrome?', <u>Twin Research</u> <u>and Human Genetics 2019; vol.22(5), pp.343-344</u>
- French, T., Korbely, R., Armstrong, R., Gunn, J., Gallacher, L., Zhao, T., Lunke, S., Chong, B., Stark, Z. and Tan, T. [Department of Neonatology, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Rapid exome sequencing of DNA from multiple tissues identifies KRAS mosaicism in oculoectodermal syndrome', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.373</u>
- **Garland, S.** [Infection and Immunity Group, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'A significant measure of HPV vaccine effectiveness in a high-risk population in Korea prior to a National Immunization Program', <u>Journal of Gynecologic</u> <u>Oncology 2020; vol.31(1), pp.e32</u>
- Gayden, T., Sepulveda, F.E., Khuong-Quang, D.A., Pratt, J., Valera, E.T., Garrigue, A., Kelso, S., Sicheri, F., Mikael, L.G., Hamel, N., Bajic, A., Dali, R., Deshmukh, S., Dervovic, D., Schramek, D., Guerin, F., Taipale, M., Nikbakht, H., Majewski, J., Moshous, D.,

Charlebois, J., Abish, S., Bole-Feysot, C., Nitschke, P., Bader-Meunier, B., Mitchell, D., Thieblemont, C., Battistella, M., Gravel, S., Nguyen, V.H., Conyers, R., Diana, J.S., McCormack, C., Prince, H.M., Besnard, M., Blanche, S., Ekert, P.G., Fraitag, S., Foulkes, W.D., Fischer, A., Neven, B., Michonneau, D., de Saint Basile, G. and Jabado, N. [Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Author correction: germline *HAVCR2* mutations altering TIM-3 characterize subcutaneous panniculitis-like T cell lymphomas with hemophagocytic lymphohistiocytic syndrome', *Nature Genetics 2019; vol.51(1), pp.196* 

- Giouzeppos, O., Flowers, N., Shi, G., Hunt, C., Scarff, K., Archibald, A., Norris, F. and Pertile, M. [Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Post-zygotic origin of trisomy as a cause of false-negative NIPT results', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.366</u>
- **Golshevsky, D.M., Magnussen, C., Juonala, M., Kao, K.T., Harcourt, B.E. and Sabin, M.A.** [Department of General Medicine, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Centre for Hormone Research, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Time spent watching television impacts on body mass index in youth with obesity, but only in those with shortest sleep duration', *Journal of Paediatrics and Child Health 2019; Dec 10 [Epub ahead of print]*
- **Gyngell, C., Stark, Z. and Savulescu, J.** [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Drugs, genes and screens: the ethics of preventing and treating spinal muscular atrophy', <u>Bioethics 2019;</u> <u>Nov 26 [Epub ahead of print]</u>
- Haas, M.A., Boughtwood, T.F. and Quinn, M.C.J. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'The ethics approval process for multisite research studies in Australia: changes sought by the Australian Genomics initiative', <u>Medical Journal of</u> <u>Australia 2019; vol.211(10), pp.440-444.e1</u>
- Hanna, B., Stark, Z., Lunke, S., Christodoulou, J., Lewis, K., Boggs, K. and Luig, M. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Ultra-rapid genomic sequencing in the neonatal intensive care: expanding the TK2-phenotype and informing clinical decision', <u>Twin Research and Human Genetics 2019</u>; vol.22(5), pp.374
- Hedrick, K., Armstrong, G. and Borschmann, R. [Centre for Adolescent Health, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Self-harm among asylum seekers in Australian immigration detention', <u>Lancet Public Health 2019; vol.4(12), pp.e604</u>
- Heerde, J.A., Bailey, J.A., Toumbourou, J.W. and Catalano, R.F. [Department of Paediatrics, University of Melbourne; Population Health Studies of Adolescents, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Longitudinal associations between the adolescent family environment and young adult substance use in Australia and the United States', <u>Frontiers in Psychiatry 2019; vol.10, 821</u>
- Hernstadt, H., Cheung, A., Hurem, D., Vasilunas, N., Phuong, L.K., Quinn, P., Agrawal, R.,
   Daley, A.J., Cole, T. and Gwee, A. [Department of General Medicine, Department of Infectious Diseases, Department of Immunology, Department of Microbiology, Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Changing epidemiology and predisposing factors for invasive pneumococcal disease at two Australian tertiary hospitals', *Pediatric Infectious Disease Journal 2020; vol.39(1), pp.1-6*
- Heron, S., Gardner, A., Corbett, M., Bennett, M., Regan, B., Grinton, B., Coleman, M., Hildebrand, M., Bahlo, M., Scheffer, I., Gecz, J. and Berkovic, S. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Vesicular GABA co-transporter variants are associated with genetic epilepsy with febrile seizures plus (GEFS+)', *Twin Research and Human Genetics 2019; vol.22(5), pp.341*
- Hiam, D., Voisin, S., Denham, J., Yan, X., Landen, S., Jacques, M., Garnham, A. and Eynon,N. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Telomere length in

skeletal muscles and leukocytes, and aerobic fitness', *<u>Twin Research and Human Genetics</u>* 2019; vol.22(5), pp.409

- Hiam, D., Voisin, S., Denham, J., Yan, X., Landen, S., Jacques, M., Garnham, A. and Eynon,
   N. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Telomere length in skeletal muscle and leukocytes, and aerobic fitness', *Twin Research and Human Genetics* 2019; vol.22(5), pp.358
- Hoq, M., Ali, M., Islam, A. and Banerjee, C. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Risk factors of acute malnutrition among children aged 6-59 months enrolled in a community-based programme in Kurigram, Bangladesh: a mixed-method matched casecontrol study', *Journal of Health, Population and Nutrition 2019; vol.38(1), pp.36*
- Hossny, E., Ebisawa, M., El-Gamal, Y., Arasi, S., Dahdah, L., El-Owaidy, R., Galvan, C.A., Lee,
   B.W., Levin, M., Martinez, S., Pawankar, R., Tang, M.L.K., Tham, E.H. and Fiocchi, A.
   [Department of Allergy and Immunology, Royal Children's Hospital; Murdoch Childrens
   Research Institute; Department of Paediatrics, University of Melbourne, Flemington
   Road, Parkville, Vic., Australia.], 'Challenges of managing food allergy in the developing
   world', *World Allergy Organization Journal 2019; vol.12(11), article no. 100089*
- Hui, L. and Bianchi, D.W. [Reproductive Epidemiology Group, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Fetal fraction and noninvasive prenatal testing: what clinicians need to know', <u>Prenatal Diagnosis 2019; Dec 10 [Epub ahead of print]</u>
- Hui, L., Lindquist, A., Poulton, A., Kluckow, E., Hutchinson, B., Bonacquisto, L., Pertile,
  M.D., Gugasyan, L., Kulkarni, A., Harraway, J., Howden, A., McCoy, R., da Silva Costa,
  F., Palma-Dias, R., Nisbet, D., Martin, N., Behune, M., Poulakis, Z. and Halliday, J.
  [Reproductive Epidemiology, Prevention Innovation Group, Murdoch Childrens Research
  Institute; Department of Paediatrics, University of Melbourne; Victorian Infant Hearing
  Screening Program, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.],
  'State-wide performance of traditional and cell-free DNA-based prenatal testing
  pathways: the Victorian Perinatal Record Linkage (PeRL) study', <u>Ultrasound in Obstetrics</u>
  and Gynecology 2019; vol.54(Suppl. 1), pp.1-2
- Jarmolowicz, A., Brett, G., West, K., Smagarinsky, Y., Valente, G., Allen, E., Prawer, Y., MacCiocca, I. and Lynch, E. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'One size doesn't fit all. Genetic counselor reflections of implementing genomic sequencing', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.393</u>
- Kanga-Parabia, A., Kennedy, J., Gaffney, L., Davis, T. and Fawcett, S. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Experiences of women and their partners receiving variants of uncertain significance through prenatal chromosomal microarray', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.393-394</u>
- King, G., Winlaw, D.S., Alphonso, N., Andrews, D., Finucance, K., Konstantinov, I.E. and d'Udekem, Y. [Department of Cardiac Surgery, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Heart Research Group, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Atrioventricular valve closure in Fontan palliation', *European Journal of Cardio-Thoracic Surgery 2019; [Epub ahead of print]*
- Koay, H.F., Su, S., Amann-Zalcenstein, D., Daley, S.R., Comerford, I., Miosge, L., Whyte, C.E., Konstantinov, I.E., d'Udekem, Y., Baldwin, T., Hickey, P.F., Berzins, S.P., Mak, J.Y.W., Sontani, Y., Roots, C.M., Sidwell, T., Kallies, A., Chen, Z., Nussing, S., Kedzierska, K., Mackay, L.K., McColl, S.R., Deenick, E.K., Fairlie, D.P., McCluskey, J., Goodnow, C.C., Ritchie, M.E., Belz, G.T., Naik, S.H., Pellicci, D.G. and Godfrey, D.I. [Royal Children's Hospital; Melbourne Children's Centre for Cardiovascular Genomics and Regenerative Medicine, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'A divergent transcriptional landscape underpins the development and functional branching of MAIT cells', <u>Science Immunology 2019; vol.4(41), article no. eaay6039 (full text via Document Delivery)</u>

- Lee, M.G.Y., Mynard, J.P., Luitingh, T.L., Walker, A.M., Cheung, M.M.H., Konstantinov, I.E., Brizard, C.P., Lambert, G.W. and d'Udekem, Y. [Department of Cardiac Surgery, Department of Cardiology, Department of Nephrology, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Heart Research, Clinical Sciences, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Major device-dependence of measured hypertensive status from 24-hour ambulatory blood pressure monitoring after aortic coarctation repair', <u>Heart, Lung and Circulation 2019;</u> vol.28(7), pp.1082-1089
- Li, A.D., Bellis, E.K., Girling, J.E., Jayasinghe, Y.L., Grover, S.R., Marino, J.L. and Peate, M. [Department of Paediatric and Adolescent Gynaecology, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Centre for Adolescent Health, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Unmet needs and experiences of adolescent girls with heavy menstrual bleeding and dysmenorrhea: a qualitative study', *Journal of Pediatric and Adolescent Gynecology 2019; [Epub ahead of print]*
- Little, M.H. and Lawlor, K.T. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Recreating, expanding and using nephron progenitor populations', <u>Nature Reviews. Nephrology 2019; Dec 6 [Epub</u> <u>ahead of print]</u>
- Low, Y., McKnight, C., Kang, Y., Jackson, T., Hock, D., Stroud, D., Elliot, D., Hanssen, E., Ryan, M., Stojanovski, D., Thorburn, D. and Frazier, A. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Modelling the mitochondrial disease sengers syndrome using human embryonic stem cells', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.400</u>
- Low, Y., McKnight, C., Kang, Y., Jackson, T., Hock, D., Stroud, D., Elliot, D., Hanssen, E., Ryan, M., Stojanovski, D., Thorburn, D. and Frazier, A. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Modeling the mitochondrial disease sengers syndrome using human embryonic stem cells', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.356</u>
- Lynch, E., Maher, F., Martyn, M., Nisselle, A., Charles, T., McEwan, C. and Gaff, C. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Genomics in clinical care: preparing non-genetic health professionals', <u>Twin Research and Human Genetics 2019;</u> vol.22(5), pp.411
- Malalasekera, V., Soosay Raj, T., Routledge, D., Hill, A., Marino, P., Badman, J., Mechinaud,
   F. and Harrison, S. [Children's Cancer Centre, Royal Children's Hospital, Flemington
   Road, Parkville, Vic., Australia.], 'Extracorporeal photopheresis for pediatric patients with
   graft-vs-host disease after haematopoietic stem cell transplantation: an Australian
   experience', *HemaSphere 2019; vol.3(Suppl. 1), pp.1046-1047*
- Martin, C.A., Papadopoulos, N., Rinehart, N. and Sciberras, E. [Health Services, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Associations between child sleep problems and maternal mental health in children with ADHD', <u>Behavioral Sleep Medicine 2019; Nov 25 [Epub ahead</u> of print] (full text via Document Delivery)
- Massie, R.J. [Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Repurposing medical equipment', *Medical Journal of Australia 2019; vol.211(11), pp.527-528*
- Mawad, N., Trieu, L., Murthi, A., Smith, M., Smagarinsky, Y., Ayres, S., McGregor, C., Bruno, D., Massie, J. and Greaves, R. [Murdoch Childrens Research Institute, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Victorian newborn screening expanded CFTR mutation panel (2007-2018)', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.415</u>
- McCallum, Z., Nisselle, A., McClaren, B., Curnow, L., Smagarinsky, Y., Savoia, H., Amor, D. and White, S.M. [Royal Children's Hospital; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Game of genomes: the exomes are coming:

what do pediatricians need to know?', *Twin Research and Human Genetics 2019; vol.22(5),* pp.375-376

- McGillivray, G., Chan, F., Yeung, A., Vasudevan, A., Stark, Z., Prystupa, S., Chan, Y., Leong, T., Ireland-Jenkins, K., Fawcett, S., Graetz, M., Rose, K., Ayres, S., Jarmolowicz, A., Brett, G., Prower, Y., Chalinor, H., Dao, C., Davis, T., Hui, L., Teoh, M., Rowlands, S., Walker, S., Lynch, E., Martyn, M., Chong, B., Gaff, C., Lunke, S. and Collet, J.
   [Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Exome sequencing enhances the diagnostic rate of perinatal autopsy: a prospective multicenter clinical utility trial with implications for prenatal diagnosis', *Twin Research and Human Genetics 2019; vol.22(5), pp.340*
- McKay, E., Richmond, S., Kirk, H., Anderson, V., Catroppa, C. and Cornish, K. [Clinical Sciences, Murdoch Childrens Research Institute; Psychology Service, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Training attention in children with acquired brain injury: a study protocol of a randomised controlled trial of the TALI attention training programme', <u>BMJ Open 2019; vol.9(12), pp.e032619</u>
- Middleton, A., Milne, R., Howard, H., Niemiec, E., Robarts, L., Critchley, C., Nicol, D.,
   Prainsack, B., Atutornu, J., Vears, D.F., Smith, J., Steed, C., Bevan, P., Scott, E.R., Bobe,
   J., Goodhand, P., Kleiderman, E., Thorogood, A. and Morley, K.I. [Murdoch Childrens
   Research Institute, Parkville, Vic., Australia.], 'Members of the public in the USA, UK, Canada
   and Australia expressing genetic exceptionalism say they are more willing to donate
   genomic data', *European Journal of Human Genetics 2019; [Epub ahead of print]*
- Milgrom, Z., Amor, D., Kalro, K. and Lanyon, K. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Consumer driven genetic testing: a new model of genetic counseling for the genomic ERA', <u>Twin Research and Human Genetics 2019; vol.21(5)</u>, <u>pp.474-475</u>
- Missen, S., Wilson, C., Reed, P., Roxburgh, R., Rodrigues, M., Poke, G., Robertson, S., Potter, H., Murphy, R., Thorburn, D. and Glamuzina, E. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Mitochondrial disease in New Zealand: a nationwide prevalence study', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.345</u>
- Mould, E.V., Lau, L., Arndt, G., Barahona, P., Cowley, M.J., Ekert, P., Failes, T., Fletcher, J., Gifford, A., Grebert-Wade, D., Haber, M., Kamili, A., Kumar, A., Lock, R.B., Marshall, G.M., Mayoh, C., Norris, M., O'Brien, T., Quang, D.A.K., Strong, P., Sherstyuk, A., Trahair, T., Tsoli, M., Tucker, K., Warby, M., Wong, M., Xie, J., Ziegler, D.S. and Tyrrell, V. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Zero childhood cancer: a comprehensive precision medicine platform for children with high-risk cancer', *Cancer Research 2019; vol.79(13 Suppl.), Abstract 3111*
- Mudge, A.J., Sangeux, M., Wojciechowski, E.A., Louey, M.G., McKay, M.J., Baldwin, J.N., Dwan, L.N., Axt, M.W. and Burns, J. [Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Can pedobarography predict the occurrence of heel rocker in children with lower limb spasticity?', <u>Clinical</u> <u>Biomechanics 2020; vol.71, pp.208-213</u>
- Nabais Sa, M.J., Venselaar, H., Wiel, L., Trimouille, A., Lasseaux, E., Naudion, S., Lacombe, D., Piton, A., Vincent-Delorme, C., Zweier, C., Reis, A., Trollmann, R., Ruiz, A., Gabau, E., Vetro, A., Guerrini, R., Bakhtiari, S., Kruer, M.C., Amor, D.J., Cooper, M.S., Bijlsma, E.K., Barakat, T.S., van Dooren, M.F., van Slegtenhorst, M., Pfundt, R., Gilissen, C., Willemsen, M.A., de Vries, B.B.A., de Brouwer, A.P.M. and Koolen, D.A. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Department of Neurodevelopment & Disability, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy', *Genetics in Medicine 2019; [Epub ahead of print]*
- Nisselle, A., Martyn, M., Jordan, H., Kaunein, N., McEwen, A., Patel, C., Terrill, B., Bishop, M., Metcalfe, S. and Gaff, C. [Department of Paediatrics, University of Melbourne; Genomics in Society, Murdoch Childrens Research Institute, Parkville, Vic., Australia.],

'Ensuring best practice in genomic education and evaluation: a program logic approach', *<u>Frontiers in Genetics 2019; vol.10, 1057</u>* 

- Nisselle, A., McClaren, B., King, E., Crellin, E., Metcalfe, S. and Gaff, C. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'The changing nature of genomic medicine: Australian medical specialists' current practice, preparedness and preferences', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.335</u>
- Notini, L., Vasileva, D., Orchanian-Cheff, A. and Buchman, D.Z. [Biomedical Ethics Research Group, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Ethical issues associated with solid organ transplantation and substance use: a scoping review', <u>Monash Bioethics Review 2019; vol.37(3-4), pp.111-135 (full text via Document Delivery)</u>
- Nugraha, R., Kamath, S.D., Johnston, E., Karnaneedi, S., Ruethers, T. and Lopata, A.L. [Centre for Food and Allergy Research, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Conservation analysis of B-cell allergen epitopes to predict clinical crossreactivity between shellfish and inhalant invertebrate allergens', *Erontiers in Immunology* 2019; vol.10, 2676
- **O'Rourke, K.M., Yelland, J., Newton, M. and Shafiei, T.** [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'An Australian doula program for socially disadvantaged women: developing realist evaluation theories', <u>Women and Birth 2019:</u> <u>Nov 24 [Epub ahead of print] (full text via Document Delivery)</u>
- Papadopoulos, N., Sciberras, E., Hiscock, H., Williams, K., McGillivray, J., Mihalopoulos, C., Engel, L., Fuller-Tyszkiewicz, M., Bellows, S.T., Marks, D., Howlin, P. and Rinehart, N. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Sleeping sound with autism spectrum disorder (ASD): study protocol for an efficacy randomised controlled trial of a tailored brief behavioural sleep intervention for ASD', <u>BMJ Open 2019; vol.9(11), pp.e029767</u>
- Perrett, K.P., Halperin, S.A., Nolan, T., Carmona Martinez, A., Martinon-Torres, F., Garcia-Sicilia, J., Virta, M., Vanderkooi, O.G., Zuccotti, G.V., Manzoni, P., Kostanyan, L., Meyer, N., Ceregido, M.A., Cheuvart, B., Kuriyakose, S.O., Stranak, Z., Merino Arribas, J.M., Cilleruelo Ortega, M.J., Miranda-Valdivieso, M., Arias Novas, B., Ramos Amador, J.T., Omenaca, F., Baca, M., Marchisio, P.G. and Mesaros, N. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Impact of tetanus-diphtheriaacellular pertussis immunization during pregnancy on subsequent infant immunization seroresponses: follow-up from a large randomized placebo-controlled trial', <u>Vaccine</u> <u>2019; [Epub ahead of print]</u>
- Perrett, K.P., Halperin, S.A., Nolan, T., Martinez Pancorbo, C., Tapiero, B., Martinon-Torres, F., Stranak, Z., Virta, M., Vanderkooi, O.G., Kosina, P., Encinas Pardilla, M.B., Cristobal Garcia, I., Zuccotti, G.V., Kostanyan, L., Meyer, N., Ceregido, M.A., Cheuvart, B., Kuriyakose, S.O., Marcos Fernandez, M., Rodriguez Zambrano, M.A., Martin Garcia, A., Asenjo de la Fuente, J.E., Camacho Marin, M.D., de la Calle Fernandez-Miranda, M., Romero Espinar, Y., Marchisio, P.G., Manzoni, P. and Mesaros, N. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Immunogenicity, transplacental transfer of pertussis antibodies and safety following pertussis immunization during pregnancy: evidence from a randomized, placebocontrolled trial', *Vaccine 2019; [Epub ahead of print]*
- **Peters, H.** [Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'The potential of mRNA therapy for inborn errors of metabolism including MMA', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.363-364</u>
- **Pflaumer, A.** [Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Long-term follow-up of epicardial pacing and LV dysfunction in children with congenital heart block (Commentary)', <u>Annals of Thoracic Surgery 2019; Nov 28 [Epub ahead of print]</u>
- Phillips, N., Dalziel, S.R., Borland, M.L., Dalton, S., Lyttle, M.D., Bressan, S., Oakley, E., Hearps, S.J.C., Kochar, A., Furyk, J., Cheek, J.A., Gilhotra, Y., Neutze, J. and Babl, F.E. [Emergency Research Unit, Murdoch Childrens Research Institute; Department of

Paediatrics, University of Melbourne; Emergency Department, Royal Children's Hospital, Flemington Road, Parkville. Vic., Australia.], 'Imaging and admission practices in paediatric head injury across emergency departments in Australia and New Zealand: a PREDICT study', <u>Emergency Medicine Australasia 2019; [Epub ahead of print]</u>

- Pittet, L.F., Abbas, M., Siegrist, C.A. and Pittet, D. [Infectious Diseases Unit, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Missed vaccinations and critical care admission: all you may wish to know or rediscover-a narrative review', <u>Intensive Care</u> <u>Medicine 2019; Nov 26 [Epub ahead of print]</u>
- **Pope-Couston, R. and Delatycki, M.** [Bruce Lefroy Centre for Genetic Health Research, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Changes in perception of my role as a genetic counselor throughout a difficult case', *<u>Twin Research and Human</u> <u>Genetics 2019; vol.22(5), pp.360</u>*
- **Quinlan, C.** [Department of Nephrology, Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'CUBN variants uncouple proteinuria from kidney function', <u>Nature Reviews. Nephrology 2019; Dec 13 [Epub ahead of print]</u>
- Rafehi, H., Szmulewicz, D., Bennett, M., Sobreira, N., Pope, K., Smith, K., Gillies, G.,
  Diakumis, P., Dolzhenko, E., Eberle, M., Garcia Barcina, M., Breen, D., Chancellor, A.,
  Cremer, P., Delatycki, M., Fogel, B., Hackett, A., Halmagyi, M., Kapetanovic, S., Lang,
  A., Mossman, S., Mu, W., Patrikios, P., Perlman, S., Rosemargy, I., Storey, E.,
  Watson, S., Wilson, M., Zee, D., Valle, D., Amor, D., Bahlo, M. and Lockhart, P. [Bruce
  Lefroy Centre, Murdoch Childrens Research Institute; Department of Paediatrics,
  University of Melbourne, Parkville, Vic., Australia.], 'Homozygous inheritance of AAGGG
  RE in RFC1 causes CANVAS', *Twin Research and Human Genetics 2019; vol.22(5), pp.336-337*
- Richmond, C., Campbell, S., Foo, H.W., Lunke, S., Stark, Z., Bannister, E., Greenway, A. and Brown, N. [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute; Department of Gastroenterology, Department of Haematology, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Rapid neonatal diagnosis of biallelic SPTB mutation causing severe hemolytic anemia with liver failure', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.376</u>
- Riggs, E., Kilpatrick, N., Slack-Smith, L., Chadwick, B., Yelland, J., Muthu, M.S. and Gomersall, J.C. [Department of Paediatrics, University of Melbourne; Vascular Biology, Murdoch Childrens Research Institute; Plastic and Maxillofacial Surgery, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Interventions with pregnant women, new mothers and other primary caregivers for preventing early childhood caries', <u>Cochrane Database of Systematic Reviews 2019; Issue 11, Art. no. CD012155</u>
- Riley, L., Rudinger-Thirion, J., Frugier, M., Wilson, M., Kevin, L., Nixon, C., Kirk, E., Roscioli, T., Lunke, S., Stark, Z., Wierenga, K., Palle, S., Walsh, M., Higgs, E., Arbuckle, S., Thirukeswaran, S., Compton, A., Thorburn, D. and Christodoulou, J. [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'The expanding LARS2 pheno typic spectrum: HLASA, Perrault syndrome with leukodystrophy, and mitochondrial myopathy', *Twin Research and Human Genetics 2019; vol.22(5), pp.402*
- Ritchie, A., Tan, T., Chong, B., White, S., Stark, Z. and Lunke, S. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Improving the efficiency of variant prioritization', *Twin Research and Human Genetics 2019; vol.22(5), pp.367*
- Rius, R., Van Bergen, N.J., Compton, A.G., Riley, L.G., Kava, M.P., Balasubramaniam, S., Amor, D.J., Fanjul-Fernandez, M., Cowley, M.J., Fahey, M.C., Koenig, M.K., Enns, G.M., Sadedin, S., Wilson, M.J., Tan, T.Y., Thorburn, D.R. and Christodoulou, J. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Clinical spectrum and functional consequences associated with bi-allelic pathogenic PNPT1 variants', *Journal of Clinical Medicine 2019; November [Epub ahead of print]*

- Sadedin, S., Lunke, S. and Leng, C. [Murdoch Childrens Research Institute,Parkville, Vic., Australia.], 'Challenges and benefits of migrating to the new genome build', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.369</u>
- Sakowski, S.A., Cameron, F. and Feldman, E.L. [Department of Endocrinology and Diabetes and Centre for Hormone Research, Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Metabolism, cognition, and the brain throughout life', <u>Neurobiology of Disease 2020; vol.134, article no. 104698</u>
- Savarirayan, R. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Drug trials in skeletal dysplasias', *Twin Research and Human Genetics 2019; vol.22(5), pp.333-334*
- Schultz, J., Edwards, T., Ruddle, J., Hewitt, A., Kearns, L., McCombe, M., James, P., Winship, I. and Huq, A. [Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Implementation of a dedicated ocular genetics clinic: a clinical audit', <u>Twin Research and</u> <u>Human Genetics 2019; vol.22(5), pp.396</u>
- Senn, E., Symeonides, C., Vuillermin, P. and Ponsonby, A.L. [Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Early life microbial exposure, child neurocognition and behaviour at 2 years of age: a birth cohort study', *Journal of Paediatrics and Child Health 2019; Nov 29 [Epub ahead of print]*
- **Seymour, M., Giallo, R. and Wood, C.E.** [Intergenerational Health Population Health, Murdoch Childrens Research Institute; Royal Children's Hospital; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Perceptions of social support: comparisons between fathers of children with autism spectrum disorder and fathers of children without developmental disabilities', *Journal of Intellectual Disability Research 2019; Dec 5 [Epub ahead of print] (full text via Document Delivery)*
- Shanthikumar, S., Steinfort, D.P. and Ranganathan, S. [Department of Respiratory and Sleep Medicine, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Respiratory Diseases, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Interventional bronchoscopy in children: planning the path ahead', <u>Pediatric Pulmonology 2019; Dec 9 [Epub ahead of print]</u>
- Shern Lee, W., Stephenson, S., Freytag, S., Owens, H., Richards, K., Petrou, S., Barton, S., MacGregor, D., D'Arcy, C., Maixner, W., Harvey, A., Bahlo, M., Leventer, R. and Lockhart, P. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Department of Neurology, Department of Pathology, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Genetic and cellular characterization of cortical dysplasia using patient-derived brain tissue', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.337</u>
- Shilling, H., Murray, G., Brotherton, J.M.L., Hawkes, D., Saville, M., Sivertsen, T., Chambers, I., Roberts, J., Farnsworth, A., Garland, S.M., Hocking, J.S., Kaldor, J., Guy, R., Atchison, S., Costa, A.M., Molano, M. and Machalek, D.A. [Murdoch Childrens Research Institute; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Monitoring human papillomavirus prevalence among young Australian women undergoing routine chlamydia screening', <u>Vaccine 2019; [Epub ahead of print]</u>
- Sinclair, A. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Human sex reversal is caused by duplication or deletion of core enhancers upstream of SOX9', <u>Twin Research and Human Genetics 2019</u>; <u>vol.22(5), pp.338</u>
- Smagarinsky, Y., MacCiocca, I., White, S., Savoia, H., Phillips, J., Marcus, P., Leventer, R. and Curnow, L. [Royal Children's Hospital; Victorian Clinical Genetics Services, Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Evidence, education and encouragement - implementation of a laboratory-based genetic counselor', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.396</u>
- Snow, K.J., Cruz, A.T., Seddon, J.A., Ferrand, R.A., Chiang, S.S., Hughes, J.A., Kampmann, B., Graham, S.M., Dood, P.J., Houben, R.M., Denholm, J.T., Sawyer, S.M. and Kranzer, K. [Murdoch Childrens Research Institute; Department of Paediatrics, University of

Melbourne; Centre for Adolescent Health, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Adolescent tuberculosis', *Lancet Child and Adolescent Health* 2020; vol.4(1), pp.68-79

- Stutterd, C., Brock, S., Stouffs, K., Lockhart, P.J., Pope, K., McGillivray, G., Mandelstam, S., Jansen, A. and Leventer, R.J. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Genetic analysis of polymicrogyria in cohort of 124 patients using a deep sequencing gene panel', <u>Twin Research and Human Genetics 2019; vol.22(5)</u>, pp.377
- Sung, V., Downie, L., Paxton, G., Rose, E., Peadon, E., Liddle, K., Roddick, L., Chan, W., Cottier, C., Harris, A., Hunter, M., Peacock, K., Saunders, K. and Amor, D. [Victorian Clinical Genetics Service, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Childhood hearing Australasian Medical Professionals (CHAMP) network: consensus guidelines recommendations on etiological investigation and clinical medical management of childhood hearing loss in Australia', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.372</u>
- Sweeney, R., Chen, G., Gold, L., Mensah, F. and Wake, M. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Mapping PedsQL<sup>™</sup> scores onto CHU9D utility scores: estimation, validation and a comparison of alternative instrument versions', <u>Quality of Life Research 2019; [Epub ahead</u> <u>of print]</u>
- Tan, N., Stapleton, R., Stark, Z., White, S., Delatycki, M., Yeung, A., Hunter, M., Amor, D., Brown, N., Stutterd, C., McGillivray, G., Yap, P., Regan, M., Chong, B., Fanjul
   Fernandez, M., Marum, J., Phelan, D., Lunke, S. and Tan, T. [Victorian Clinical Genetics Services, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'What to do with a negative exome?', *Twin Research and Human Genetics 2019; vol.22(5), pp.340-341*
- Tang, M.L.K., Lozinsky, A.C. and Loke, P. [Allergy Immunology, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne; Department of Allergy and Immunology, Royal Children's Hospital, Flemington Road, Parkville, Vic., Australia.], 'Peanut oral immunotherapy: state of the art', <u>Immunology and Allergy Clinics of North</u> <u>America 2020; vol.40(1), pp.97-110</u>
- Thorburn, D., Pantaleo, S., Smith, M., Akesson, L., Christodoulou, J., Du Sart, D. and Chong, B. [Victorian Clinical Genetic Services, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'The role of prenatal diagnosis in prevention of mitochondrial DNA disease', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.366</u>
- Thorne, N., Lunke, S., Fellowes, A., Martyn, M., Nisselle, A., Roesley, A., McEwen, C., Fanjul Fernandez, M., Tan, T., Liddicoat, D., Stark, Z., Thompson, E., Maher, F., Cunningham, F., MacCiocca, I., Reid, G., Jame, P., Hodgson, J. and Gaff, C. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Variant interpretation training for the genomics era: a 5-year comprehensive strategy aiming towards professional competencies', *Twin Research and Human Genetics 2019; vol.22(5), pp.368*
- Thorne, N., Lunke, S., Fellowes, A., Martyn, M., Nisselle, A., Roesley, A., McEwen, C., Fanjul Fernandez, M., Tan, T., Liddicoat, D., Stark, Z., Thompson, E., Maher, F., Cunningham, F., MacCiocca, I., Reid, G., Jame, P., Hodgson, J. and Gaff, C. [Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Variant interpretations training for the genomics era: a 5-year comprehensive strategy aiming towards professional competencies', *Twin Research and Human Genetics 2019; vol.22(5), pp.415*
- Toh, Z.Q., Kosasih, J., Russell, F.M., Reyburn, R., Fong, J., Tuivaga, E., Ratu, F.T., Nguyen, C.D., Matanitobua, S., Do, L.A.H., Menheniott, T., Frazer, I.H., Garland, S.M., Mulholland, E.K. and Licciardi, P.V. [Infection and Immunity, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic.,

Australia.], 'Selective persistence of HPV cross-neutralising antibodies following reduceddose HPV vaccine schedules', *Vaccine 2019; [Epub ahead of print]* 

- Tosif, S., Jatobatu, A., Maepioh, A., Subhi, R., Francis, K.L. and Duke, T. [Centre for International Child Health, Clinical Epidemiology and Biostatistics Unit, Murdoch Childrens Research Institute; Royal Children's Hospital; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Cause-specific neonatal morbidity and mortality in the Solomon Islands: an assessment of data from four hospitals over a three-year period', *Journal of Paediatrics and Child Health 2019; Dec 10 [Epub ahead of print]*
- Turk, B.R., Theda, C., Fatemi, A. and Moser, A.B. [Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'X-linked adrenoleukodystrophy: pathology, pathophysiology, diagnostic testing, newborn screening, and therapies', <u>International Journal of Developmental Neuroscience 2019; Nov</u> 25 [Epub ahead of print]
- Uebergang, E., De Silva, M., Best, S. and Finlay, K. [Department of Paediatrics, University of Melbourne; Victorian Clinical Genetics Services, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Helping culturally diverse populations understand genomics: development and evaluation of a plain language resource', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.416</u>
- Vasiljevski, E., Houweling, P., Summers, M., Little, D. and Schindeler, A. [Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Treating muscle weakness and fatigue in neurofibromatosis type 1', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.416-417</u>
- Vears, D., Senecal, K. and Borry, P. [Biomedical Ethics Research Group, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Exploration of interactions between genetic health professionals and laboratory specialists in clinical genomic sequencing', <u>Twin</u> <u>Research and Human Genetics 2019; vol.22(5), pp.341</u>
- Velasco-Arnaiz, E., Soriano-Arandes, A., Espiau, M., Altet, N., Fortuny, C., Monsonis, M., Tebruegge, M. and Noguera-Julian, A. [Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Impact of baseline tuberculin skin test and isoniazid chemoprophylaxis on subsequent quantiferon-Tb gold in-tube performance in young children assessed after tuberculosis contact in catalonia', <u>Pediatric Infectious</u> <u>Disease Journal 2020; vol.39(2), pp.e22-e25</u>
- Wallis, M., Rafehi, H., Hildebrand, M.S., Valente, G., Murdoch, E., Uebergang, E., Chalinor, H., Stutterd, C., Huq, A., Salmon, L., Eratne, D., Mullen, S., White, S., Brown, N., Christodoulou, J., Bahlo, M., Berkovic, S.F. and Delatycki, M. [Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Closing a gap for adults with rare and undiagnosed conditions: the Austin Health Adult Undiagnosed Diseases Program (AHA-UDP)', <u>Twin Research and Human Genetics 2019; vol.22(5), pp.377</u>
- Wheeler, B.J., Snoddy, A.M.E., Munns, C., Simm, P., Siafarikas, A. and Jefferies, C. [Department of Endocrinology and Diabetes, Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'A brief history of nutritional rickets', <u>Frontiers</u> <u>in Endocrinology 2019; vol.10, 795</u>
- Wiedemann, A., Pastore-Wapp, M., Slavova, N., Steiner, L., Weisstanner, C., Steinlin, M., Grunt, S., Mori, A.C., Bigi, S., Datta, A., Fluss, J., Hackenberg, A., Keller, E., MacKay, M.T., Maier, O., Mercati, D., Marcoz, J.P., Poloni, C., Ramelli, G.P., Regenyi, M., Schmid, R. and Schmitt-Mechelke, T. [Department of Paediatrics, Royal Children's Hospital; Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Impact of stroke volume on motor outcome in neonatal arterial ischemic stroke', <u>European Journal of Paediatric</u> <u>Neurology 2019; [Epub ahead of print]</u>

- Yeung, A., Tan, N., Tan, T., Stark, Z., Brown, N., Delatycki, M., Hunter, M., Lunke, S., Chong, B., Phelan, D., Brett, G., Jarmolwicz, A., Prawer, Y., Martyn, M., McEwan, C., Gaff, C. and White, S. [Victorian Clinical Genetics Services, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'Evaluating cost-effectiveness of exome sequencing in a prospective versus historical cohort of complex pediatric patients', <u>Twin Research and Human Genetics 2019; vol.22(5)</u>, <u>pp.338</u>
- Yigit, G., Saida, K., DeMarzo, D., Miyake, N., Fujita, A., Yang Tan, T., White, S.M., Wadley, A., Toliat, M.R., Motameny, S., Franitza, M., Stutterd, C.A., Fee Chong, P., Kira, R., Sengoku, T., Ogata, K., Guillen Sacoto, M.J., Fresen, C., Beck, B.B., Nurnberg, P., Dieterich, C., Wollnik, B., Matsumoto, N. and Altmuller, J. [Victorian Clinical Genetics Services, Murdoch Childrens Research Institute; Department of Paediatrics, University of Melbourne, Parkville, Vic., Australia.], 'The recurrent post-zygotic pathogenic variant p.Glu47Lys in RHOA causes a novel recognizable neuro-ectodermal phenotype', <u>Human Mutation 2019; Dec 10 [Epub ahead of print]</u>
- Yong, M.S., Zhu, M.Z.L., Huang, L., Griffiths, S., Brink, J., Brizard, C.P., d'Udekem, Y. and Konstantinov, I.E. [Department of Cardiac Surgery, Royal Children's Hospital; Department of Paediatrics, University of Melbourne; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Long-term quality of life outcomes in adult survivors after anomalous pulmonary venous drainage repair', <u>Annals of Thoracic Surgery 2019; Nov 30 [Epub ahead of print]</u>
- Young, J.T. and Hughes, N. [Centre for Adolescent Health, Murdoch Childrens Research Institute, Parkville, Vic., Australia.], 'Traumatic brain injury and homelessness: from prevalence to prevention', *Lancet Public Health 2019; Dec 2 [Epub ahead of print]*
- Zhang, T., Wallis, M., Petrovic, V., Challis, J., Kalitsis, P. and Hudson, D.F. [Cytogenetics Department, Victorian Clinical Genetics Services, Murdoch Childrens Research Institute; Royal Children's Hospital; Department of Paediatrics, University of Melbourne, Flemington Road, Parkville, Vic., Australia.], 'Loss of TOP3B leads to increased R-loop formation and genome instability', *Open Biology 2019; vol.9(12), article no. 190222*
- Zou, Z., Li, C. and Patton, G.C. [Department of Paediatrics, University of Melbourne; Centre for Adolescent Health, Royal Children's Hospital; Murdoch Childrens Research Institute, Flemington Road, Parkville, Vic., Australia.], 'Early-life exposure to the Chinese famine and subsequent T2DM', <u>Nature Reviews. Endocrinology 2019; [Epub ahead of print]</u>

#### BOOKS

#### [online]



to quickly learn the basics.

Before we are born: essentials of embryology and birth defects / Keith L. Moore, T.V.N. Persaud and Mark G. Torchia. 10<sup>th</sup> ed. Elsevier, 2020. [<u>8<sup>th</sup> ed, 2013</u> held in print]

As the study of embryology continues to be integrated with a range of disciplines, Before We Are Born remains the ideal solution for students who need

Fully updated by the world's foremost embryologists, this medical reference book provides concise guidance on human embryology at every stage of development, utilizing rich illustrations and photographs designed to further explain content.

#### [online]



<u>Clinical examination skills in</u> <u>paediatrics: for MRCPCH candidates</u> <u>and other practitioners</u> / edited by A. Mark Dalzell, Ian Sinha. Wiley-

Blackwell, 2020. Examining children presents unique

challenges for trainees and new doctors in paediatrics. Paediatric patients vary greatly in age and

development, often find it difficult to describe their symptoms, and can behave unpredictably in clinical settings.

Clinical Examination Skills in Paediatrics helps MRCPCH candidates and other practitioners learn effective history taking and fundamental examination techniques.

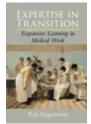
Clear and concise chapters–with contributions from a team of paediatric specialists–demonstrate the clinical

examination and questioning techniques used in daily practice.

Emphasis on the intellectual processes involved in decision making assists both trainees preparing for a formal examination as well as new clinicians faced with a difficult diagnostic problem.

Topics include cardiovascular and respiratory examination, examining a child with a neuromuscular disorder, musculoskeletal examination, and taking history from a child with diabetes and a rheumatological condition.

#### [in print] 153.9 ENG



Expertise in transition: expansive learning in medical work / Yrjo Engestrom. Cambridge University Press, 2018

This book challenges standard notions of expertise. In today's world, truly effective expertise is built on fluid

collaboration between practitioners from multiple backgrounds.

Such collaborative expertise must also be transformative, must be able to tackle emerging new problems and changes in its organizational framework.

Engeström argues that the transition toward collaborative and transformative expertise is based on three pillars: expertise needs to be understood and cultivated as a collective activity; expertise needs to be built on flexible knot-working among diverse practitioners; and expertise needs to be fostered as the expansive learning of models and patterns of activity that are in progress.

In this book, Engeström recasts expertise as fluid collaboration on complex tasks that requires envisioning the future and mastering change.

#### [in print] 616.8 FEN [online]



#### Fenichel's Clinical pediatric neurology: a signs and symptoms approach / [edited by] J. Eric Pina-Garza, Kaitlin C. James. 8th ed.

Elsevier, 2019

Provides a solid foundation in the diagnosis and management of primary neurologic disorders of childhood using step-by-step, authoritative guidance that considers each presenting symptom in terms of differential diagnosis and treatment, reflecting real-life patient evaluation and management.

Key features include: a new chapter on genetics, and weighted differential diagnosis tables and treatment algorithms that help you quickly identify the more common and most treatable neurological disorders. This book also brings you up to date with the new definition of status epilepticus; new guidelines for Lennox Gastaut syndrome; new FDA-approved drugs for epilepsy, ADHD, dystonia, and more; new data on sudden infant death syndrome; and revised consensus criteria which unifies the concepts of neuromyelitis optica (NMO) and neuromyelitis optica spectrum disorders (NMOSD).

#### [online]



#### Multidisciplinary management of pediatric voice and swallowing disorders / J. Scott McMurray, Matthew R. Hoffman, Maia N. Braden, editors. Springer, 2020.

This text provides a comprehensive review of the assessment and

management of pediatric voice and swallow disorders from the perspectives of both the pediatric laryngologist as well as the speech-language pathologist whose collaboration is critical to effective clinical care.

All chapters are written by experts in dual fields and formatted to present a straightforward approach to diagnosing and managing each disorder, including descriptions of relevant operative interventions.

Multiple intraoperative photographs and illustrations depicting how to perform each surgical procedure are also included.

This text will serve as a useful step-by-step guide and resource not only for otolaryngologists and speechlanguage pathologists, but all members of the pediatric aerodigestive team and other providers caring for children affected by voice and swallowing disorders.

#### [online]



<u>Pediatric hand therapy</u> / edited by Joshua M. Abzug, Scott H. Kozin, and Rebecca Neiduski. Elsevier, 2020.

Get a quick, expert overview of optimal therapeutic interventions for pediatric patients with musculoskeletal

conditions of the hand.

This concise resource by Drs. Joshua Abzug, Scott Kozin, and Rebecca Neiduski offers practical recommendations and guidelines along with key background information, for a well-rounded, concise perspective on hand therapy for children.

You'll benefit from the knowledge and experience of orthopaedic surgeons and other members of the rehabilitation care team who provide a real-world, multidisciplinary approach to this complex field.

#### [online]



Severe asthma in children and adolescents: mechanisms and

<u>management</u> / Erick Forno, Sejal Saglani, editors. Springer, 2020.

This book provides readers with a comprehensive review of severe asthma in children and adolescents, covering

epidemiology, genetics, risk factors, co-morbidities, clinical presentation, diagnosis and treatment.

Written by leaders in the field, chapter discussions draw on the mechanisms driving the disease, genetics, multidisciplinary approaches, immunomodulators, and other important aspects in the management of the disease.

It is designed to be a practical guide, opening with an introduction on the epidemiology of severe childhood asthma, as well as a discussion of special considerations of the disease unique to the preschool-aged patient.

The distinctions between the diagnosis and management of asthma in young children of various ages, and specific approaches for the adolescent with severe asthma, including the transition into adult care are then addressed.

Finally, the book closes with a discussion on the current state and future avenues for severe asthma research. Severe Asthma in Children and Adolescents is an indispensable reference for the healthcare professional, for basic and translational researchers, as well as for students, residents and fellows.

#### [online]



Transition from pediatric to adult healthcare services for adolescents and young adults with long-term conditions: an international perspective on nurses' roles and interventions / Cecily L. Betz, Imelda

T. Coyne, editors. Springer, 2020.

This book offers essential information on interventions and actions that enable and promote transition experiences for adolescents and young adults.

It provides guidance on appropriate strategies that bring together these groups and caregivers in the context of transition preparation, and those which optimize adolescents' and young adults' ability to self-manage their healthcare.

The health care transition for adolescents and young adults has gained more attention given the improved survival rates, including for those with long-term conditions. It is now estimated that more than 90% of children diagnosed with a long-term condition will survive into adulthood. The significant change in survival requires concomitant services to facilitate adolescents' and young adults' successful transition to adult health care, therefore providers need education, knowledge and skills to support healthcare transition services.

A recent US national survey demonstrated only 10% of parents/caregivers reported that their children received transition preparation services. This book is targeted at nurses and pediatric and adult health care providers of different disciplines seeking guidance on which interventions are available, how they can be used, advantages and challenges, and how best to promote a seamless transition for all adolescents and young adults with long-term conditions.

It provides several examples of transition programmes and initiatives worldwide. There is an increasing focus on how nurses can support transition but little guidance on what is effective and what has been tried. This book will fill a gap by addressing all of these issues outlined above and by providing worked examples from leading nurse researchers and academics worldwide.