

Paediatrician, Rehabilitation Medicine Paediatrician

2010-Current

The Children's Hospital at Westmead, cnr Hawkesbury and Hainsworth street, Westmead NSW 2145 Australia.

I provide interdisciplinary care to children and adolescents with congenital and acquired limb loss and disabling musculoskeletal conditions (skeletal dysplasia, rare Ehlers-Danlos syndromes, Arthrogryposis, Osteogenesis Imperfecta and Marfan). I also provide care for children with neurological disability including brain and spinal cord injury.

2011-Current

Narrabeen Sports Medicine Centre, Sydney Academy of Sport, Wakehurst Parkway, Narrabeen NSW 2101 Australia

My private practice includes children with complex pain, disabling fatigue, musculoskeletal conditions, difficulties with motor skills and Joint Hypermobility (Hypermobility Spectrum Disorder and hEDS).

Qualifications:

FRACP	Fellowship of the Royal Australian College of Physicians (RACP) (Paediatrics) 2009
FAFRM	Fellowship of the Australasian Faculty of Rehabilitation Medicine (AFRM), RACP 2008
MSPMed	Masters in Sports Medicine, University of New South Wales, Australia 2006
MRCPCH	Membership of the Royal College of Paediatrics and Child Health (RCPCH), UK, 2001
DCH	Diploma in Child Health, RCPCH 2000, UK
MBBS	Bachelor of Medicine, Bachelor of Surgery St Bartholomew's and the Royal London Hospitals, University of London, UK 1998
BSc	Bachelor of Science with First Class Honours, University of London, UK 1995

Professional Accreditations and Organisations

AHPRA: Full National General and Specialist (Paediatrics, Rehabilitation) Medical Registration (MED0001194095)
General Medical Council UK: Full registration (4517405)
Fellow, Australasian Faculty of Rehabilitation Medicine
Fellow, Royal Australasian College of Physicians
Member, Royal College of Paediatrics and Child Health (UK)
Member, International Society of Prosthetics and Orthotics (ISPO)
Chair, Paediatric working group of the Ehlers-Danlos Society International
Member, hEDS/HSD committee the Ehlers-Danlos Society International
Member, Sports Medicine Australia
Medical Advisory Board, Short Statured people of Australia

Invited International Speaker

2021 International Society of Prosthetics and Orthotics (ISPO) World congress: Paediatric Upper Limb Symposium, 2nd November 2021 (UK/ Australia collaboration)
2019 ICCBH invited expert "Expert round table - management of foramen magnum stenosis in infants with Achondroplasia" Salzburg, Austria June 2019.
2018 Biomarin Pharmaceuticals "Achondroplasia in Australia: growth and neurocognitive profiles" Achondroplasia Natural history expert meeting
2017 ISPO world congress, Cape Town, South Africa. Symposium on "Management and outcomes for children with congenital and acquired limb loss" (with the SCHN limb clinics teams)
2016 London Hypermobility Masterclass (95% positive feedback)

Invited National Speaker:

2020 Australian College of Sports Physicians Annual Conference

“Hypermobility in Children: What’s normal and who needs to be referred”
2018 Ehlers Danlos International Society Professionals day and Patient day Sydney and Perth
2017 Human Genetics Society Australia 41st Annual Scientific Meeting, Association of Clinical Geneticists satellite.
“What do we do with all these hypermobile kids”
2014 Spring Meeting of the Faculty of Pain Medicine, Australian and New Zealand College of Anaesthetists
Invited speaker on Joint Hypermobility Syndrome with Verity Pacey
21st Annual Scientific Meeting of the Australasian Faculty of Rehabilitation Medicine Sydney 2013
Concurrent session: “Sprains Strain and Pain: Growing Pains”
Closing plenary: Hypermobility – To move or not to move (with Dr Tan, A/Prof Nicholson and Verity Pacey)

Research Activity

H- Index 10 (scopus accessed Jan 2022)

ORCID: [https://orcid.org/ 0000-0002-3961-0493](https://orcid.org/0000-0002-3961-0493)

Sapphire: RES-ID-1141363

Current projects:

“Diagnostic Criteria for Children with Hypermobility Disorders” for Ehlers-Danlos International
“Changes in physical function over 3 years of young persons with longitudinal fibular deficiency” data collection
Principal Investigator, “Vosoritide for Achondroplasia” (BMN 111-302 and 111-206 follow on studies active)
“Podiatric management for children with symptomatic joint hypermobility” PhD programme (MM)
“Health and Ability of children with Achondroplasia” (management guidelines, medical complications, physical function and caregiver projects underway)
“The clinical characteristics of children with joint hypermobility and their relationship to adverse outcomes” 5-year cohort study. data collection complete, 5 full publications, 2 further papers in draft.

Scholarships and Grants:

2017 RACP project grant \$10, 000 (for limb projects)

2014 Zimmer grant \$10, 000 (for hypermobility projects)

2012 Australian Rheumatology Association project grant \$ 15, 000 (for hypermobility projects)

2006 Clancy-Donald fellowship \$ 30,000 for a 12-month clinical sports medicine fellowship

University Affiliation – Macquarie University Conjoint Lecturer/HDR supervisor (2020-ongoing)

Student Supervision:

University of Sydney Summer students (Honours projects)

2010 Sandeep Das (Exercise Physiology) “Growth Charts for Children with Achondroplasia”

2015 Emma Seigal (Psychology) “Depression and Anxiety in Children with Hypermobility”

Macquarie University Masters by Research (clinical co-supervision)

2018 Joshua Pate (Physiotherapy) complete

2019 Elle Morris (Physiotherapy) complete

2020 Jennifer Armstrong (Physiotherapy) complete

University of Sydney Doctor of Philosophy (clinical supervision, one year only)

2014 Verity Pacey (Physiotherapy) complete

University of Newcastle Doctor of Philosophy

2020 Muhammed Maarj (Podiatry) current (clinical co-supervisor)

Papers in peer reviewed journals:

Maarj, M., Coda, A., **Tofts, L.**, Santos, D., Pacey, V. Outcome measures for assessing change over time in studies of symptomatic children with hypermobility: a systematic review *BMC Pediatrics* 2021, 21(1), 527

Ravi Savarirayan, **Louise Tofts**, Melita Irving, William Wilcox, Carlos A Bacino, Julie Hoover-Fong *et al* Safe and persistent growth-promoting effects of vosoritide in children with achondroplasia: 2-year results from an open-label, phase 3 extension study. *Genetics in Medicine* 23(12) pg2443-2447.

Ireland, P.J., Savarirayan, R., Pocovi, T., Tate, T., Coussens, M., **Tofts, L.**, Munns, C., Pacey, V. Development of the Screening Tool for Everyday Mobility and Symptoms (STEMS) for skeletal dysplasia (2021) *Orphanet Journal of Rare Diseases*, 16 (1) n. 40.

Hoover-Fong, J., Cheung, M.S., Fano, V., Hagenas, L., Hecht, J.T., Ireland, P., Irving, M., Mohnike, K., Offiah, A.C., Okenfuss, E., Ozono, K., Raggio, C., **Tofts, L.**, Kelly, D., Shediach, R., Pan, W., Savarirayan, R. Lifetime impact of achondroplasia: Current evidence and perspectives on the natural history (2021) *Bone*, 146, n. 115872.

Tofts, L., Armstrong, J., Pacey, Childhood prevalence of achondroplasia in New South Wales and the Australian Capital Territory, Australia (2021) *American Journal of Medical Genetics, Part A*, 185 (5), pp. 1481-1485.

Eleanor J. Morris, **Louise Tofts**, Margaret Patterson, Oliver Birke, Roger Adams, Adrienne Epps, Kathrine Knox, Marnee J. McKay, Jennifer N. Baldwin, Joshua Burns & Verity Pacey (2020): Physical performance of children with longitudinal fibular deficiency (fibular hemimelia), *Disability and Rehabilitation*.

Ravi Savarirayan, **Louise Tofts**, Melita Irving, William Wilcox, Carlos A Bacino, Julie Hoover-Fong *et al* Once-Daily subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial. *Lancet* 2020, 396: 684-692.

Pate, J.W.; Hancock, M.J.; **Tofts, L.**; Epps, A.; Baldwin, J.N.; McKay, M.J.; Burns, J.; Morris, E.; Pacey, V. Longitudinal Fibular Deficiency: A Cross-Sectional Study Comparing Lower Limb Function of Children and Young People with That of Unaffected Peers. *Children* **2019**, 6, 45.

Lesley L Nicholson, Roger D. Adams, Louise **Tofts** and Verity Pacey (2017). Physical and Psychosocial Characteristics of Current Child Dancers and Nondancers With Systemic Joint Hypermobility: A Descriptive Analysis. *J Orthop Sports Phys Ther* 2017;47(10):782-791.

Louise **Tofts**, Sandeep Das, Felicity Collins and Karen L.O. Burton (2017). Growth Charts for Australian children with Achondroplasia. *Am J Med Genet. Part A* 173 (8), pp. 2189-2200

Mark C. Scheper, Lesley L Nicholson, Roger D. Adams, Louise **Tofts** and Verity Pacey (2017). The natural history of children with joint hypermobility syndrome and Ehlers Danlos Hypermobility type: a longitudinal cohort study. *Rheumatology* 56 (12), pp. 2073-2083.

Scheper MC, Pacey V, Rombaut L, Adams R, **Tofts L**, Calders P, Nicholson LL and Engelbert RHH (2017). Generalised Hyperalgesia in children and adults diagnosed with Ehlers Danlos Hypermobility type: A discriminative analysis. *Arthritis Care and Research* 69 (3), pp. 421-429.

Wigg, Kimberly, **Tofts Louise**, Benson Suzanne and Porter Melanie (2016). The Neuropsychological function of Children with Achondroplasia. *American Journal of Medical Genetics* 170 (11), pp. 2882-2888.

Pacey, V, **Tofts, L**, Adams, R., Munns, C., Nicholson, L (2015). Quality of Life prediction in children with joint hypermobility syndrome. *J. Paed Child Health*. 51(7), 689-95.

Pacey, V, **Tofts, L**, Wesley A, Collins FC and Singh-Grewal D (2015). Joint hypermobility syndrome: a review for clinicians. *J. Paed Child Health*. 51(4), 373-80.

Pacey, V, **Tofts, L**, Adams, R., Munns, C., Nicholson, L. (2015), Joint Hypermobility Syndrome subclassification in paediatrics: a factor analytical approach. *Arch Dis Child* 100(1) 8-13.

Pacey, V, **Tofts, L**, Adams, R., Munns, C., Nicholson, L (2014). Proprioceptive acuity into knee hypermobile range in children with joint hypermobility syndrome. *Pediatric Rheumatology Online Journal*. 12(8), 40.

Tofts LJ and Hamblin N (2014). C-Leg improves function and quality of life in an adolescent traumatic trans femoral amputee – a case study. *Prosthetics and Orthotics International*. 38(5), 413-417.

Pacey, V, **Tofts, L**, Adams, R., Munns, C., Nicholson, L (2013). Exercise in children with joint hypermobility syndrome and knee pain: a randomised controlled trial comparing exercise into hypermobile versus neutral knee extension. *Pediatric Rheumatology Online Journal*. 11(1), 30.

Tofts LJ, Elliott EJ, Munns C, Pacey V, Sillence DO. The differential diagnosis of children with joint hypermobility: a review of the literature. *Paediatric Rheumatology*. 2009 Jan 5;7:1.

Tofts, LJ. Stanley, CS. Barnett, TG and Logan, JG. Knee joint function and the energy cost of level walking in soccer players. *British Journal of Sports Medicine*. 1998; 32(2):130-133.

Presented posters and abstracts:

Jennifer Armstrong, Verity Pacey and **Louise Tofts** (2021).

A Retrospective Cohort study of Medical Complications in an Australian Population of Children with Achondroplasia
Childhood Prevalence of achondroplasia in New South Wales and the ACT, Australia
Human Genetics Society Australia 44st Annual Scientific Meeting, 2021

Louise Tofts “What do we do with all these hypermobile kids” platform presentation. *Human Genetics Society Australia 41st Annual Scientific Meeting, Association of Clinical Geneticists satellite meeting 2017*

Louise Tofts, Emma Siegal, Roger Adams, Leslie Nicholson and Verity Pacey. Anxiety and Depression in Australian children and adolescents with Joint Hypermobility Syndrome.

Verity Pacey, **Louise Tofts**, Roger Adams and Leslie Nicholson. Quality of life of children with JHS/EDS-HT: who will decline over 18 months.

Ehlers-Danlos Society 2016 International Symposium, New York.

Pacey, V, **Tofts L**, Adams R and Nicholson L. (2015) Factors affecting change in children with joint hypermobility syndrome: results of a prospective longitudinal study. *Internal Medicine Journal* 2015; 45 9-9

Nicholson, L., Pacey, V., **Tofts, L.**, Munns, C., Adams, R. (2014). Signs and symptoms of foot and ankle dysfunction in children with joint hypermobility. *Journal of Foot and Ankle Research*, 7 (Suppl 1).

Pacey, V, **Tofts L**, Adams R and Nicholson L. Comparison of quality of life of children with Joint Hypermobility Syndrome, Charcot-Marie-Tooth disease and the general Australian paediatric population. *Abstract 8*

Pacey, V, **Tofts L**, Adams R, Munns C and Nicholson L. A RCT comparing two different exercise programmes for children with joint hypermobility syndrome (JHS) and knee pain. *Abstract 10.*

21st Annual Scientific Meeting of the Australasian Faculty of Rehabilitation Medicine, 2013

Pacey, V, **Tofts L**, Adams R, Munns C and Nicholson L. The fatigue experience of children with joint hypermobility syndrome: A child and parent perspective. *Internal Medicine Journal* 2013; 43 (suppl. 2) 7-8 RHPA 5 *54th Annual Scientific Meeting of the Australian Rheumatology Association, 2013*

Pacey, V, **Tofts L**, Adams R, Nicholson L and Munns C. Exercise Programmes for children with joint hypermobility syndrome and knee pain: a randomised controlled trial. *Internal Medicine Journal* 2012; 42 (suppl. 1) 36-38 RHPA P1 *54th Annual Scientific Meeting of the Australian Rheumatology Association, 2012*

S Das, F Collins and **LJ Tofts**. Head circumference for age curves in Australian boys with Achondroplasia and the association between head circumference and cervicomedullary compression.

S Das, F Collins and **LJ Tofts**. Descriptive Height, Weight and BMI for age charts in Australian boys with achondroplasia.

Posters at the 10th Meeting of the International Skeletal Dysplasia Society 2011

Paget SP and **Tofts LJ**. Growth plate injury due to meningiococcal disease in childhood: a cause of delayed limb deformity impacting on orthopaedic and prosthetic management. (*poster*)

Hamblin N and **Tofts L**. C-Leg improves function and quality of life in an adolescent traumatic transfemoral amputee (*platform*)

Tofts L and Farid M. The role of antenatal counselling at the time of diagnosis of congenital limb loss. (*poster*)

Presented at the Annual meeting of the International Society of Prosthetics and Orthotics Australia 2011

LJ Tofts, EJ Elliott, C Munns, V Pacey and DO Sillence. The differential diagnosis of children with joint hypermobility: a review of the literature *Paediatric Rheumatology* 2008, **6**(Suppl 1):P156

LJ Tofts, S Hayden, MC Waugh. Effect of Botulinum Toxin type-A (Botox®) on neck pain and craniocervical headaches caused by trapezius spasm in a child with generalised joint hypermobility resulting from Noonan's Syndrome.

Paediatric Rheumatology 2008, **6**(Suppl 1):P174

Posters at the Paediatric Rheumatology European Society meeting, London UK 2008

T.G. Barnett, J.G. Logan, Clare. S. Stanley and **Louise Tofts**. Energy cost of walking in soccer players. *J. Physiol.* 1995; **487**: 63P *Poster at the physiological society meeting, Cork, Ireland 1995*