

Dr Fiona Connell's publications

Mutations in GATA2 cause primary lymphedema associated with a predisposition to acute myeloid leukemia (Emberger syndrome).

Ostergaard P, Simpson MA, Connell FC, Steward CG, Brice G, Woollard WJ, Dafou D, Kilo T, Smithson S, Lunt P, Murday VA, Hodgson S, Keenan R, Pilz DT, Martinez-Corral I, Makinen T, Mortimer PS, Jeffery S, Trembath RC, Mansour S. *Nat Genet.* 2011 Sep 4;43(10):929-31

Rapid identification of mutations in GJC2 in four limb primary lymphoedema using whole exome sequencing combined with linkage analysis

Pia Ostergaard, Michael A Simpson, Glen Brice, Sahar Mansour, Fiona Connell, Alexandros Onoufriadis, Anne H Child, Jae Hwang, Kamini Kalidas, Peter Mortimer, Richard Trembath, Steve Jeffery. *J Med Genet.* 2011 Apr;48(4):251-5. Epub 2011 Jan 25.

Emberger syndrome-primary lymphedema with myelodysplasia: report of seven new cases.

Mansour S, Connell F, Steward C, Ostergaard P, Brice G, Smithson S, Lunt P, Jeffery S, Dokal I, Vulliamy T, Gibson B, Hodgson S, Cottrell S, Kiely L, Tinworth L, Kalidas K, Mufti G, Cornish J, Keenan R, Mortimer P, Murday V; Lymphoedema Research Consortium. *Am J Med Genet A.* 2010 Sep;152A(9):2287-96

Re: "conjunctival edema and distichiasis in association with congenital lymphedema of the lower legs".

Brice G, Connell F, Mansour S, Jeffery S, Mortimer P. *Ophthal Plast Reconstr Surg.* 2010 May-Jun;26(3):222-3

A new classification system for primary lymphatic dysplasias based on phenotype.

Connell F, Brice G, Jeffery S, Keeley V, Mortimer P, Mansour S. *Clin Genet.* 2010 May;77(5):438-52

Milroy Disease.

Brice GW, Mansour S, Ostergaard P, Connell F, Jeffery S, Mortimer P. In: Pagon RA, Bird TC, Dolan CR, Stephens K, editors. *GeneReviews* [Internet]. Seattle (WA): University of Washington, Seattle; 1993. 2006 Apr 27 [updated 2009 Jul 23]

Linkage and sequence analysis indicate that CCBE1 is mutated in recessively inherited generalised lymphatic dysplasia.

Connell F, Kalidas K, Ostergaard P, Brice G, Homfray T, Roberts L, Bunyan DJ, Mitton S, Mansour S, Mortimer P, Jeffery S; Lymphoedema Consortium. *Hum Genet.* 2010 Feb;127(2):231-41. Epub 2009 Nov 13. Erratum in: *Hum Genet.* 2010 Feb;127(2):243

Analysis of the coding regions of VEGFR3 and VEGFC in Milroy disease and other primary lymphoedemas.

Connell FC, Ostergaard P, Carver C, Brice G, Williams N, Mansour S, Mortimer PS, Jeffery S; Lymphoedema Consortium.
Hum Genet. 2009 Jan;124(6):625-31. Epub 2008 Nov 12. Erratum in: Hum Genet. 2009 Mar;125(2):237

Congenital vascular malformations: a series of five prenatally diagnosed cases.

Connell F, Homfray T, Thilaganathan B, Bhide A, Jeffrey I, Hutt R, Mortimer P, Mansour S.

Am J Med Genet A. 2008 Oct 15;146A(20):2673-80

Phenotypic characterization of primary lymphedema.

Connell F, Brice G, Mortimer P.

Ann N Y Acad Sci. 2008;1131:140-6. Review