New cause identified for children and adults with joint, skeletal and skin problems

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Scientists from The University of Manchester and Central Manchester University Hospitals NHS Foundation Trust have identified the cause of a rare condition called Leri's pleonosteosis (LP).

Dr Sid Banka from the Manchester Centre for Genomic Medicine at The University of Manchester, led a team of researchers on the study which is published in *Annals of Rheumatic Diseases* journal on 18th January 2014.

LP is an inherited condition in which children are born with contractures of multiple joints and then develop difficulty of joint movements that progress in severity with age. The research team showed that extra genetic material on chromosome number 8 caused the condition in two families from Manchester.

Some patients with LP also develop thickening of their skin, similar to that seen in patients with a more common disorder called scleroderma.

Using their new knowledge, the research team showed that the genetic cause of LP is linked to whether people get scleroderma or not.

This work opens opportunities to understand scleroderma and explore new treatments.

More information: "Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing GDF6

and SDC2 and provides insight into systemic sclerosis pathogenesis." Siddharth Banka, Stuart A Cain, Sabrya Carim, Sarah B Daly, Jill E Urquhart, Günhan Erdem, Jade Harris, Michelle Bottomley, Dian Donnai, Bronwyn Kerr, Helen Kingston, Andreas Superti-Furga, Sheila Unger, Holly Ennis, Jane Worthington, Ariane L Herrick, Catherine L R Merry, Wyatt W Yue, Cay M Kielty, William G Newman. *Ann Rheum Dis Annrheumdis*-2013-204309Published Online First: 17 January 2014 DOI: 10.1136/annrheumdis-2013-204309