

## Publications for David Sillence

### 2009

Ramjan, K., Roscioli, T., Rutsch, F., Sillence, D., Munns, C. Generalized arterial calcification of infancy: treatment with bisphosphonates. *Nature Clinical Practice Endocrinology & Metabolism*. 2009; 5:167-172.

Tofts, L., Elliott, E., Munns, C., Pacey, V., Sillence, D. The differential diagnosis of children with joint hypermobility: a review of the literature. *Pediatric Rheumatology Online Journal*. 2009; 7:1.

### 2008

Kaplan, F., Xu, M., Glaser, D., Collins, F., Connor, M., Kitterman, J., Sillence, D., Zackai, E., Ravitsky, V., Zasloff, M., Ganguly, A., Shore, E. Early diagnosis of fibrodysplasia ossificans progressiva. *Pediatrics*. 2008; 121:e1295-300.

Gleeson, H., Wiltshire, E., Briody, J., Hall, J., Chaitow, J., Sillence, D., Cowell, C., Munns, C. Childhood chronic recurrent multifocal osteomyelitis: pamidronate therapy decreases pain and improves vertebral shape. *The Journal of Rheumatology*. 2008; 35:707-712.

### 2006

Gabbett, M., Jones, K., Cowell, C., Sillence, D., Wilson, M. Neonatal severe hyperparathyroidism: An important clue to the aetiology. *Journal of paediatrics and child health*. 2006; 42:813-6.

Sparrow, D., Chapman, G., Wouters, M., Whittock, N., Ellard, S., Fatkin, D., Turnpenny, P., Kusumi, K., Sillence, D., Dunwoodie, S. Mutation of the LUNATIC FRINGE gene in humans causes spondylocostal dysostosis with a severe vertebral phenotype. *American journal of human genetics*. 2006; 78:28-37.

### 2005

Bajaj, R., Smith, J., Trochet, D., Pitkin, J., Ouvrier, R., Graf, N., Sillence, D., Kluckow, M. Congenital central hypoventilation syndrome and Hirschsprung's disease in an extremely preterm infant. *Pediatrics*. 2005; 115:e737-8.

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Fleming, F., Woodhead, H., Briody, J., Hall, J., Cowell, C., Ault, J., Kozlowski, K., Sillence, D. Cyclic bisphosphonate therapy in osteogenesis imperfecta type V. *Journal of paediatrics and child health*. 2005; 41:147-51.

Grewal, S., Wynn, R., Abdenur, J., Burton, B., Gharib, M., Haase, C., Hayashi, R., Shenoy, S., Sillence, D., Tiller, G., Dudek, M., van Royen-Kerkhof, A., Wraith, J., Woodard, P., Young, G., Wulfraat, N., Whitley, C., Peters, C. Safety and efficacy of enzyme replacement therapy in combination with hematopoietic stem cell transplantation in Hurler syndrome. *Genetics in medicine: official journal of the American College of Medical Genetics*. 2005; 7:143-6.

Neas, K., Smith, J., Chia, N., Huseyin, S., St Heaps, L., Peters,

G., Sholler, G., Tzioumi, D., Sillence, D., Mowat, D. Three patients with terminal deletions within the subtelomeric region of chromosome 9q. *American Journal of Medical Genetics. Part A*. 2005; 132:425-30.

### 2004

David, G., Sillence, D., Hardwick, R., Opitz, J. A case of Kabuki (Niikawa-Kuroki) syndrome associated with manifestations resembling C-trigonocephaly syndrome. *American Journal of Medical Genetics. Part A*. 2004; 130A:389-92.

Hein, L., Bawden, M., Muller, V., Sillence, D., Hopwood, J., Brooks, D. alpha-L-iduronidase premature stop codons and potential read-through in mucopolysaccharidosis type I patients. *Journal of molecular biology*. 2004; 338:453-62.

### 2003

Robinson, C., Sillence, D. The osteodystrophy of mucopolipidosis type III and the effects of intravenous pamidronate treatment. *Journal Of Inherited Metabolic Disease*. 2003; 25:681-693.

Van Der Slott, A., Sillence, D., 13 Researchers from, P. Identification of PLOD2 as telopeptide lysyl hydroxylase, an important enzyme in fibrosis. *Journal Of Biological Chemistry*. 2003; 278 (42):40967-40972.

Lam, W., Chan, H., Sillence, D. Desbuquois syndrome: Clinical and radiological report for the first two Chinese cases from a consanguineous family. *Journal of Paediatrics and Child Health*. 2003; 39:707-712.

### 2002

Beighton, P., Francomano, C., Giedion, A., Hall, C., Hall, J., Horton, W., Kaitila, I., Krakow, D., Lachman, R., Le Merrier, M., Mortier, G., Mundlos, S., Poznanski, A., Rimoin, D., Savarirayan, R., Spranger, J., Superti-Furga, A., Unger, S., Washbrook, J., Warman, M., Wilcox, W., Winter, R., Sillence, D. International nosology and classification of constitutional disorders of bone (2001). *American Journal Of Medical Genetics*. 2002; 113:65-77.

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Cowell, C., Sillence, D., Briody, J., Hall, J., Ault, J., Hooper, M. Monthly versus second monthly intravenous pamidronate therapy for osteogenesis imperfecta. *Pediatric Endocrinology Montreal 2001*. 2001; 38(1):56-63.

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Taillandier, A., Lia-Baldini, A., Mouchard, M., Muller, F., Simon-Bouy, B., Serre, J., Bera-Louville, A., Bonduelle, M., Eckhardt, J., Gaillard, D., Myhre, A., Kortge-Jung, S., Larget-Pier, L., Malou, E., Sillence, D., Temple, I., Mornet, E. Twelve novel mutations in the tissue-nonspecific alkaline phosphatase gene (ALPL) in patients with various forms of hypophosphatasia. *Human Mutation*. 2001; 108:179-185.

Dahlstrom, J., Arbuckle, S., Kozlowski, K., Peek, M.,

Thomson, M., Reynolds, G., Sillence, D. Lethal prenatal onset infantile cortical hyperostosis (Caffey disease). *Pathology*. 2001; 37:91-93.

Al-Agha, A., Cowell, C., Briody, J., Hall, J., Anderson, D., Sillence, D. Cyclic intravenous Pamidronate therapy in chronic recurrent multifocal osteomyelitis (CRMO): A report of two cases. *Pediatric Endocrinology Montreal 2001*. 2001; 27, No. 3:309-312.

## 2000

Sillence, D., Briody, J., Hall, J., Ault, J., Howman-Giles, R., Cowell, C., Hooper, M. Cyclic intravenous pamidronate therapy for osteogenesis imperfecta. New York, NY: Elsevier Science Inc, 2000.

Rearson, W., Smith, A., Honour, J., Hindmarsh, P., Das, D., Rumsby, G., Nelson, I., Malcolm, S., Ades, L., Sillence, D., Kumar, D., DeLozier-Blanchet, C., McKee, S., Kelly, T., McKeehan, W., Baraitser, M., Winter, R. Evidence for digenic inheritance in some cases of Antley-Bixler syndrome?. *Journal of Medical Genetics*. 2000; 37:26-32.

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