Publications

Recent publications on the Axiom® Genotyping Solution

Complex disorders

Lu X., *et. al.* Genome-wide association study in Han Chinese identifies four new susceptibility loci for coronary artery disease. *Nature Genetics* **44**: 890–894 (2012).

Galanter J. M., *et. al.* A genomewide association study in 4000 Latino children identifies several candidate regions for asthma and replicates the association in 17q21.
American Journal of Respiratory and Critical Care Medicine **185**: A2504 (2012).

Davies R. W., *et. al.* A Genome Wide Association Study for Coronary Artery Disease Identifies a Novel Susceptibility Locus in the Major Histocompatibility Complex.
Circulation: Cardiovascular Genetics **5**: 217-225 (2012)

Hoffmann T. J., *et. al.* Design and coverage of high throughput genotyping arrays optimized for individuals of East Asian, African American, and Latino race/ethnicity using imputation and a novel hybrid SNP selection algorithm. *Genomics* **98**(6):422-30 (2011).

Hoffmann T. J., *et. al.* Next generation genome-wide association tool: Design and coverage of a high-throughput European-optimized SNP array. *Genomics* **98**(2):79-89 (2011).

Simple and rare disorders

Shaheen R., et. al. POC1A Truncation Mutation Causes a Ciliopathy in Humans Characterized by Primordial Dwarfism. *American Journal of Human Genetics* 91(2): 330-336 (2012).

Alshammari M. J., et. al. Mutation in RAB33B, which encodes a regulator of retrograde Golgi transport, defines a second Dyggve–Melchior–Clausen locus. *Journal of Medical Genetics* 49(7): 455-461 (2012).

Alazami A. M., et. al. Molecular characterization of Joubert syndrome in Saudi Arabia.
Human Mutation Epub. doi: 10.1002/humu.22134 (2012).

Alangari A., et. al. LPS-responsive beige-like anchor (LRBA) gene mutation in a family with inflammatory bowel disease and combined immunodeficiency.
Journal of Allergy and Clinical Immunology 130(2): 481–488 (2012).

Aldahmesh M. A., et. al. Homozygous null mutation in ODZ3 causes microphthalmia in humans.
Genetics in Medicine Epub doi:10.1038/gim.2012.71 (2012).

Khan A. O., et. al. Familial spherophakia with short stature caused by a novel homozygous ADAMTS17 mutation. *Ophthalmic Genetics* Epub doi: 10.3109/13816810.2012.666708 (2012).

Khan A. O., *et. al.* Clinical and molecular analysis of children with central pulverulent cataract from the Arabian Peninsula.
British Journal of Ophthalmology Epub doi:10.1136/bjophthalmol-2011-301053 (2012).

Al-Qattan M. M., *et. al*. Familial dorsalization of the skin of the proximal palm and the instep of the sole of the foot. *Gene* **500**(2): 216–219 (2012).

Al-Hassnan Z. N., *et. al*. Recessively Inherited Severe Aortic Aneurysm Caused by Mutated EFEMP2. *American Journal of Cardiology* **109**(11): 1677–1680 (2012).

Aldahmesh M. A., *et. al*. Identification of a truncation mutation of the acylglycerol kinase (AGK) gene in a novel autosomal recessive cataract locus. *Human Mutation* Epub doi: 10.1002/humu.22071 (2012).

Shamseldin H. E., *et. al*. Exome sequencing reveals a novel Fanconi group defined by XRCC2 mutation. *Journal of Medical Genetics* **49**:184-186 (2012)

Shamseldin H. E., *et. al*. Identification of a novel DLX5 mutation in a family with autosomal recessive split hand and foot malformation. *Journal of Medical Genetics* **49**:16-20 (2012).

Aldahmesh M. A., *et. al*. Recessive Mutations in ELOVL4 Cause Ichthyosis, Intellectual Disability, and Spastic Quadriplegia. *American Journal of Human Genetics* **89**(6):745–750 (2011).

Aldahmesh M. A., *et. al*. Novel recessive BFSP2 and PITX3 mutations: Insights into mutational mechanisms from consanguineous populations. *Genetics in Medicine* **13**:978–981 (2011).

Steinlein O. K., *et. al.* Mutations in FKBP10 can cause a severe form of isolated Osteogenesis imperfecta. *BMC Medical Genetics* **12**(1):152 epub (2011).

Shaheen R., *et. al.* A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. *Human Mutations* **32**(6):573-8 (2011).

Shaheen R., *et. al.* Recessive mutations in DOCK6, encoding the guanidine nucleotide exchange factor DOCK6, lead to abnormal actin cytoskeleton organization and Adams-Oliver syndrome. *American Journal of Human Genetics* **89**(2):328-33 (2011).

Aldahmesh M. A., *et. al.* Identification of ADAMTS18 as a gene mutated in Knobloch syndrome. *Journal of Medical Genetics* **48**(9):597-601 (2011).

Abu-Safieh, L., *et. al.* Mutation of IGFBP7 causes upregulation of BRAF/MEK/ERK pathway and familial retinal arterial macroaneurysms. *American Journal of Human Genetics* **89**(2):313-9 (2011).

Alazami A. M., *et. al.* A nullimorphic ERLIN2 mutation defines a complicated hereditary spastic paraplegia locus (SPG18). *Neurogenetics* **12**(4):333-336 (2011).

Technical papers

Hollegaard M. V., *et. al.* Robustness of genome-wide scanning using archived dried blood spot samples as a DNA source. *BMC Genetics* **12**(1):58 (2011).

Software tools and algorithms

O'Connell, J., et. al. Joint Genotype Calling With Array and Sequence Data. *Genetic Epidemiology* Epub doi: 10.1002/gepi.21657 (2012).

Seelow D., et. al. HomozygosityMapper2012--bridging the gap between homozygosity mapping and deep sequencing. *Nucleic Acids Research* **40** (W1): W516-W520 (2012).