

H3M2: Detection of runs of homozygosity from whole-exome sequencing data



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Runs of homozygosity (ROH) can be defined as sizable chromosomal stretches of homozygous genotypes, ranging in length from tens of kilobases to megabases. ROHs can be relevant for population and medical genetics, playing a role in predisposition to both rare and common disorders. ROHs are commonly detected by SNP microarrays, but attempts have been made to use Whole Exome Sequencing (WES) data. Currently available methods developed for the analysis of uniformly spaced SNP-array maps do not fit easily the sparse and non uniform distribution of the WES target design. To meet the need of an approach specifically tailored to WES data we developed H3M2, an original algorithm based on Heterogeneous Hidden Markov Model that incorporates inter-marker distances to detect ROH from Whole Exome Sequencing (WES) data. We evaluated the performance of H3M2 to correctly identify ROHs on synthetic chromosomes and examined its accuracy in detecting ROHs of different length (short, medium and long) from real 1000 genomes project data. H3M2 turned out to be more accurate than GERMLINE and PLINK, two state-of-the-art algorithms, especially in the detection of short and medium ROHs. H3M2 is freely available at <https://sourceforge.net/projects/h3m2/>.

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Whole Exome Sequencing, homozygosity, autozygosity



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Runs of homozygosity (ROH) are sizeable stretches of homozygous genotypes at consecutive polymorphic DNA marker positions, traditionally captured by means of genome-wide single nucleotide polymorphism (SNP) genotyping. Autozygosity is that particular type of homozygosity that reflects “identity by descent” (IBD) and results from the co-occurrence at a given locus of the same allele deriving from a common ancestor, by way of non-random mating (inbreeding). Long ROHs are commonly a consequence of recent parental relatedness but, in spite of their small size, even short ROHs can reflect autozygosity. With the advent of Next Generation Sequencing (NGS) technologies, a number of methods initially devised for analysis of SNP array data or specifically conceived for NGS data have been adopted for the detection of ROH from Whole Exome Sequencing (WES) data. Gene mapping strategies combining mutation detection and identification of autozygous ROH through NGS approach can be effective in several situations in which traditional mapping approaches would fail. Here we review the state of the art of gene mapping and genetic population studies focusing on IBD regions and outline future perspectives.

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Current state of biotechnology in Italy



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Despite being worldwide well known for its extraordinary natural beauty and, above all, for its historical heritage, Italy is also one of the most competitive country in the biotechnological field, thanks to 422 companies engaged in R&D in the field of biotechnologies. Among these, more than half (264) fall under the definition of pure biotech company. In recent years, biotechnologies have become a solid industrial reality in all fields of application: health, energy, agro-food and industry, in general. Despite the adverse international economic situation, in 2013 the Italian biotechnology industry has been able to make great strides internationally. Italy can be proud of being at third position in Europe, after Germany and the United Kingdom, for number of biotech companies, whose total turnover represents a more than proportional growth in investments in research and development (R&D). Active in the fields of human health, agro-food and in the industrial sector, our biotech companies confirm the ability to transform the excellence of Italian research into new products and services, and represent an extremely dynamic and competitive reality, able to overcome the cyclic nature that characterizes other industrial sectors. The number of biotech companies in Italy has sharply increased over the last decade.

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Micropatterned collagen-based scaffolds for peripheral nerve regeneration



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Nerve injury is a frequent event especially after traumatic injury, affecting mainly young people. Various therapeutic approaches have been proposed and results have been, so far, inconsistent, in terms of both quality and extent of nerve regeneration and re-innervation.

Here, a novel technique is presented able to produce tubular collagen-based scaffolds, characterized by a radially/axially patterned microporosity (MPCS). The process has been modelled by means of the Lamm differential equation, according to appropriate scaling laws.

In the present investigation, we tested in vivo clinicopathological impact of this MPCS over a 10-mm critical size defects in the adult rat sciatic nerve. Rats with transection of the sciatic nerve and implanted with either commercial collagen or silicon conduits were used as controls. MPCS-implanted rats showed significantly improved nerve regeneration at both neurophysiological and neuropathological levels, as compared to control rats. Our data demonstrate that this specific tubular scaffold micropatterning orchestrates physiological regeneration in the adult rat sciatic nerve over a 10-mm critical size defect. Indeed, whole genome gene expression analyses confirm that the MPCS induces