IntegraNews

HELPING YOU TO FIND™

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WELCOME TO IntegraNews

The goal of this newsletter is to provide an update on the latest service offers, news and events at IntegraGen Genomics.

With the addition of new partnerships with world-class academic institutions, the introduction of additional service offerings, and the availability of the latest versions of sequencing technology platforms in our lab, IntegraGen Genomics continues to be a leader in providing sequencing and bioinformatics services to researchers.

IntegraGen Genomics is also excited to announce that it will be sponsoring a satellite symposium during the **8èmes Assises de Génétique Humaine et Médicale** being held in Lyon, France in February 2016.

This symposium, entitled **Exomes and RNA-Seq: From basic science to clinical research** will feature presentations on:

- Advanced bioinformatics solutions
- The added value of exome and RNA-Seq in clinical studies
- Exome sequencing from liquid biopsies

Please join us in Lyon for these exciting presentations. For more information, visit www.integragen-genomics.com/assises-genetique-2016

Merci,

- Emmanuel Martin
  Director, Sales & Marketing
  IntegraGen Genomics

IN THIS ISSUE:

ANALYSIS OF CANCER MUTATIONAL SIGNATURES

Somatic mutations are the result of multiple mutational processes, including mutagen exposures and defective DNA repair. Different mutational processes generate unique combinations of somatic mutation types, called "mutational signatures."

In their seminal study, Alexander et al. identified 21 signatures associated with distinct types of human cancer, either associated or not associated with known etiologies (e.g. tobacco use, UV exposure, BRCA mutation)

New signatures have been identified and are reported in the Catalogue of Somatic Mutations in Cancer (COSMIC).

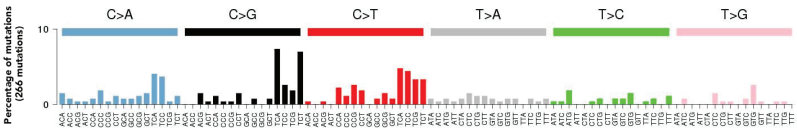

Fig 1: 96 trinucleotides mutational signature of a tumor sample from exome data.

Through its advanced genomic consulting service **GeCo**, IntegraGen Genomics offers a complete solution for mutational signatures analysis in your samples from exome or whole genome data. Our service includes:

- Detection of 96 trinucleotides signatures in each sample
- Transcriptional bias analysis
- Identification of processes occurring in a tumor series
- Comparison with known signatures listed on COSMIC
- Classification of the tumors regarding their mutational profile

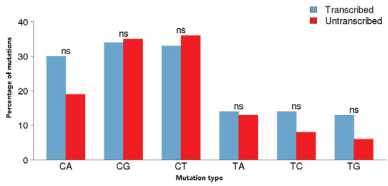

Fig 2: Transcriptional bias representation of a tumor sample

GeCo service combines cutting-edge bioinformatics and biostatistics analysis, supervised by leading research experts, to help you to get the best quality analysis from your genomic data.

Alexandrov, et al. Signatures of mutational processes in human cancer. *Nature*. 2013; 500:415–421.

INSTITUT PASTEUR ANNOUNCES ALLIANCE WITH INTEGRAGEN GENOMICS

The Institut Pasteur has announced the signing of an agreement with IntegraGen Genomics on March 24, 2015 whereby IntegraGen will be responsible for the technical and operational implementation of high-throughput sequencing activities for use by the microbiology experts of National Reference Centers (CNRs) at the Institut Pasteur.

The primary objective of the partnership between the Institut Pasteur and IntegraGen is to increase access to the latest generation of sequencing technologies for the fifteen CNRs and the Institute's microbiology collection with the additional goal of establishing reference tools for the typing of bacterial, viral, and fungal strains. The Institut Pasteur will rely on the expertise and experience of IntegraGen to help ensure optimal quality of results returned via high-throughput sequencing. IntegraGen will also help reduce delays in obtaining sequencing results and increase the flexibility of the business while optimizing the operational costs of reagents.

Additionally, IntegraGen will develop management software tailored to the internal needs of the Institut Pasteur.

Professor Christian Bréchot, President at Institut Pasteur, stated "Being at the heart of infectious disease surveillance issues worldwide, the Institute Pasteur must equip its laboratories with the best available technology. Having access to IntegraGen's know-how in high-throughput sequencing is a major asset to reinforce our mobilization abilities and to ensure our mission for public health at the highest level."

Our alliance with the Institute Pasteur further demonstrates IntegraGen Genomics' ability to provide high quality next generation sequencing services to researchers and reinforces our genomic services expertise.

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NEXT-GENERATION SEQUENCING HELPS PATIENTS WITH RETINAL DYSTROPHIES

By identifying causative mutations linked to retinal disorders, clinicians could better classify patients in sub-groups and orient them to therapeutic clinical trials.

Cone and cone-rod dystrophies are clinically and genetically heterogeneous inherited retinal disorders with predominant cone impairment. Ideally, these would be distinguishable from the more common group of rod-cone dystrophies (retinitis pigmentosa) due to their more severe visual prognosis with early central vision loss.

In collaboration with IntegraGen Genomics, the INSERM team of Institut de la Vision* uses targeted high-throughput sequencing to study 123 genes implicated in retinal diseases, in collaboration with IntegraGen Genomics. The laboratory of Dr. Christina Zeitz and Dr. Isabelle Audo identified causative mutations for 62.1% of cases, revealing 33 known and 34 novel mutations associated to autosomal dominant cases, autosomal recessive and sporadic cases. Research work is ongoing to discover new genes, by performing whole exome sequencing.

* Unit 968 Inserm/CNRS/Université Pierre et Marie Curie, Institut de la vision, Paris.
For more information visit http://goo.gl/JMRaBU

Boulanger-Scemama E *et al.* Next-generation sequencing applied to a large French cone and cone-rod dystrophy cohort: mutation spectrum and new genotype-phenotype correlation. Orphanet J Rare Dis. 2015 Jun 24;10(1):85.

RESEARCH NEWS:

IntegraGen sequencing service helps identify new virus involved in the development of liver cancer

Professor Jessica Zucman-Rossi and her colleagues at Inserm Unit 1162 in Paris identified the role of AAV2, a previously considered nonpathogenic virus, in the development of a rare type of liver cancer. The results of their studies have recently been published in *Nature Genetics*.

"Our research, which was greatly enhanced by our partnership with IntegraGen, demonstrated the involvement of AAV2 in the pathogenesis liver cancer in patients with no underlying cirrhosis and represents a new potential new etiology for this disease," stated Jessica Zucman-Rossi, M.D., Ph.D., Professor of Medical Oncology at the Paris Descartes University, Georges Pompidou European Hospital and Director of INSERM Unit 1162.

Nault, et al. Recurrent AAV2-related insertional mutagenesis in human hepatocellular carcinomas. *Nat Genet* 2015; 47:1187–1193

NEW SERVICE - OPTIMIZED PROCESS FOR MEASURING SMALL RNA EXPRESSION

IntegraGen Genomics has developed and streamlined a process for measuring small RNA expression in **tissue samples** and **liquid biopsies** (serum or plasma) via next generation sequencing. We offer researchers a small RNA sequencing service for examining gene regulation at both the transcriptional and post-transcriptional level.

SMALL RNA-SEQ

- Full optimized process – from extraction to bioinformatics
- Start with either frozen or FFPE tissue, circulating blood cells, or liquid biopsies
- Able to extract total RNA from either plasma or serum
- Library prep uses random adapters via in-house developed protocol
  - Reduces bias representation
  - Provides better correlation
- State-of-the-art analysis using snRNAbench mutational signatures

OTHER TECHNOLOGIES WE OFFER:

NANOSTRING® miRNA PANEL

- Determination of miRNA expression without amplification
- Can be used for FFPE and degraded RNA samples

RT-qPCR miRNA EXPRESSION

- RT-qPCR using Biomark™ System from Fluidigm
- Compatible with TaqMan and QIAGEN miScript primer

Better correlation with the IntegraGen Genomics protocol

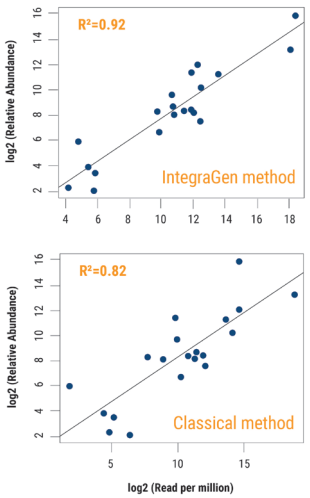

Fig 1: Correlation between qRT-PCR and NGS using IntegraGen Genomic's proprietary protocol compared to a standard protocol

Less bias with the IntegraGen Genomics protocol

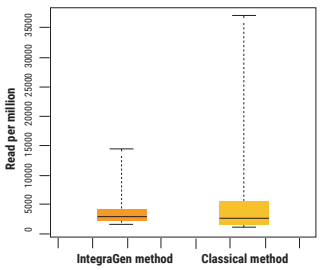

Fig 2: Distribution of the number of reads per miRNA in a reference sample (271 miRNA at the same concentration)

CONTACT US FOR A QUOTE

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At IntegraGen Genomics we have the experience and expertise to support the development and design of your genomic research projects. We are a recognized leader in the field of genomic research and provide our customers:

- Access to the latest high-throughput sequencing and molecular research technologies.
- Advanced bioinformatics and biostatistical analytical tools and services to deliver your results.
- Delivery of high-quality results in timely fashion.

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