#### **IGA Summer School 2011**

# Next Generation Sequencing and the analysis of genetic diversity

#### 6-9 September, 2011

Next Generation Sequencing (NGS) has recently revolutionized the approach to genome sequencing enabling the sequencing of billions of bases in massively parallel reactions. These new sequencing technologies collectively referred to as either 'ultra-deep' sequencing or 'massively parallel' sequencing are currently used for SNP discovery, detection of structural variants, genome-wide measurement of transcripts levels and a number of other applications, and are revolutionizing biological research.

This NGS sequencing course will take place at the Istituto di Genomica Applicata (Institute of Applied Genomics) in Udine, Italy.

It is addressed to all researchers interested in understanding how to generate sequences and interpret information obtained. It is suitable for both researchers willing to start using next generation technologies and those already working on them. The course will alternate theoretical concepts and practical examples focused on the Illumina platform. Participants are guided to follow the entire process of a next generation sequencing run, from preparing DNA/RNA libraries from the biological sample to the analysis of data.

The course is limited to a maximum of 12 participants. Applicants will be required to write a letter containing a 300-words outline describing the relevance of the course to their research.

### **Provisional Programme**

6 September 2011		Speaker	Duration	Session type
14.00	Registration			
14.30-15.00	Welcome (IGA and IGA-TS presentation)	Cattonaro	30'	Talk
15.00-16.00	Basics of NGS: comparison to conventional	Cattonaro	60'	Talk
	approaches, advantages and disadvantages of			
	different platforms			
16.00-16.15	Coffee break			
16.15-18.00	Introduction to Unix Environment	Del Fabbro	1h45'	Practical

7 September 2011				
09.00-10.00	Overview of Illumina sample preparation	Cattonaro	60'	Talk
10.00-11.00	Agilent Presentation on target resequencing (and on how to design probes for target resequencing)	Agilent	60'	Talk and pratical
11.00-11.15	Coffee break		15'	
11.15-12.00	E4 Engineering Presentation of IT solutions	E4 Computer	45'	Talk
		Engineering Spa		
12.15-13.00	Infrastructure and IT for NGS	Gervaso	60'	Talk
13.00-14.00	Lunch			
14.00-15.45	Illumina library preparation, DNA-seq, SureSelect enrichment and RNA-seq; amplify clonal clusters and perform an Illumina run	Dicenta, Felice, Jurman, Spadotto	1h45'	Practical*
15.45-16.00	Coffee Break		15'	
16.00-17.45	Illumina library preparation, DNA-seq, SureSelect enrichment and RNA-seq; amplify	Dicenta, Felice,	1h45'	Practical*

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	clonal clusters and perform an Illumina run	Jurman, Spadotto		
*Groups will be	rotating.			
8 September				
2011				
09.00–10.00	Illumina pipeline for primary data analysis and alignment to a reference	Del Fabbro	60'	Talk
10.00-11.00	Alignment to a reference	Del Fabbro	60'	Practical
11.00-11.15	Coffe break		15'	
11.15-13.00	Resequencing for variant detection: SNP,	Pinosio,	1h45'	Talk
	INDELs and structural variants	Scalabrin		
13.00-14.00	Lunch			
14.00-15.00	De novo assembly	Policriti	60'	Talk
15.00-15.45	CLC Presentation	CLC	45'	Talk
15.45-16.00	Coffee Break		15'	
16.00-17.45	CLC Workshop	CLC	1h45'	Practical
9 September				
2011				
09.00-09.45	Talk on Small RNA-seq	Horner	45'	Talk
09.45-10.30	Pooled multiplex NGS for identification of rare variants	Marroni	45'	Talk
10.30-10.45	Coffee break		15'	

45'

60'

45'

Talk

Talk

Talk

Ferrarini

Sana e

Pezzoli

Morgante

10.45 -11.30

11.30-12.30

12.30-13.15

Talk on RNA-seq

applications and perspectives

Talk on human target resequencing (pratical

experience on hypertrophic cardiomyopathy)

Conclusion talk on next generation sequencing