DEPARTMENT OF MEDICINE, HUDDINGE

H7F2537, High Throughput Functional Genomic Technologies in Biomedical Research, 1.5 credits (hec)

Storskaliga teknologier inom funktionsgenomik relevanta för biomedicinsk forskning, 1,5 högskolepoäng

Third-cycle level / Forskarnivå

Approval

This syllabus was approved by the The Committee for Doctoral Education on 2024-09-20, and is valid from spring semester 2025.

Responsible department

Department of Medicine, Huddinge, Faculty of Medicine

Prerequisite courses, or equivalent

No prerequisite courses, or equivalent, demanded for this course.

Purpose & Intended learning outcomes

Purpose

The purpose of the course is to give participants an introduction to high-throughput genomic technologies. Additionally, the participants will be able to understand which high throughput technology to apply in order to answer a specific scientific question.

Intended learning outcomes

At the end of the course the students should be able to select appropriate high throughput genomic technologies in different formats based on modern sequencing and microarray techniques, including gene expression profiling, single cell transcriptomics, ChIP sequencing, methylation profiling and other genomic methods used in modern biomedical research for different types of research questions. They should also be able to discuss advantages and disadvantages of alternative technologies.

Course content

The course includes a combination of lectures, discussions and practical sessions to gain more insight in different technological platforms used for sequencing and microarray analysis in different applications. The course includes an introduction to fundamental concepts and methods used in bioinformatics to study genome function and variation using large-scale sequencing and microarray analysis that can be applied in biomedical research including students own projects.

Forms of teaching and learning

Lectures, seminars, demonstrations and data analysis.

Language of instruction

The course is given in English

Grading scale

Pass (G) /Fail (U)

Compulsory components & forms of assessment

Compulsory components

The students have to take active part in all activities. Attendance is compulsory for demonstrations and data analysis. An alternative time could be provided only under exceptional circumstances. If it is not possible to provide an alternative time during the course, this part will need to be taken at the next course occasion. Other absence can be compensated for by an additional task in agreement with the course organizers.

Forms of assessment

Examination seminar with group presentations and discussions. The students will, in groups, select a paper of a relevant topic for the course, with the help of course leaders if necessary. The paper should be presented with specific focus on the technologies used. Each student should also be able to discuss advantages and disadvantages of alternative technologies and will be individually assessed. This seminar will take 2-3 hours.

Course literature

Recommended literature:

Reviews:
1. Trevino, V., Falciani, F., and Barrera-Saldana, H.A. (2007). DNA microarrays: a powerful genomic tool for biomedical and clinical research. Mol. Med. 13, 527-541.
2. Morozova, O., and Marra, M.A. (2008). Applications of next-generation sequencing technologies in functional genomics. Genomics 92, 255-264.

3. Mardis, E.R. (2011). A decade's perspective on DNA sequencing technology. Nature 470, 198-203.
4. Werner, T (2010). Next generation sequencing in functional genomics. Brief. Bioinform. 11, 499-511.
5. Hurd, P.J., and Nelson, C.J. (2009). Advantages of next-generation sequencing versus the microarray in epigenetic research. Brief. Funct. Genomic Proteomic 8, 174-183.
6. Jones PA. Functions of DNA methylation: islands, start sites, gene bodies and beyond. Nat Rev Genet. 2012 May 29;13(7):484-9

Reports:

1. Birney, E., Stamatoyannopoulos, J.A., Dutta, A., Guigo, R., Gingeras, T.R., Margulies, E.H., Weng, Z., Snyder, M., Dermitzakis, E.T., thurman, R.E., et al. (2007). Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature 447, 799-816.
2. Lieberman-Aiden, E., van Berkum, N.L., Williams, L., Imakaev, M., Ragoczy, T., Telling, A., Amit, I., Lajoie, B.R., Sabo, P.J., Dorschner, M.O., et al. (2009). Comprehensive mapping of long-range interactions reveals folding principles of the human genome. Science New York, NY 326, 289-293.
3. Visel, A., Blow, M.J., Li, Z., Zhang, T., Akiyama, J.A., Holt, A., Plajzer-Frick, I., Shoukry, M., Wright, C., Chen, F., et al. (2009). ChIP-seq accurately predicts tissue-specific activity of enhancers. Nature 457, 854-858.
4.Tang, F., Barbacioru, C., Wang, Y., Nordman, E., Lee, C., Xu, N., Wang, X., Bodeau, J., Tuch, B.B., Siddiqui, A., et al. (2009). mRNA-Seq whole-transcriptome analysis of a single cell. Nature methods 6, 377-382.
5. Kulasingam, V., Pavlou, M.P. and Diamandis, E.P. (2010). Integrating high-throughput technologies in the quest for effective biomarkers for ovarian cancer. Nat. Rev. Cancer 10, 371-378.
6. Zhang, J. et al. (2011). The impact of next-generation sequencing on genomics. J. Genet. Genomics 38, 95-109.
7. Macarron, R., Banks, M.N., Bojanic, D.J., Cirovic, D.A., Garyantes, T., Green, D.V.S., Hertzberg, R.P., Janzen, W.P., Paslay, J.W., Schopfer, U. and Sittampalam, G.S. (2011). Impact of high-throughput screening in biomedical research. Nat. Rev. Drug Discov. 10, 188-195.
8. Stower, H. (2012). Technology: High-throughput enhancer screening. Nat. Rev. Genet. 13, 223.