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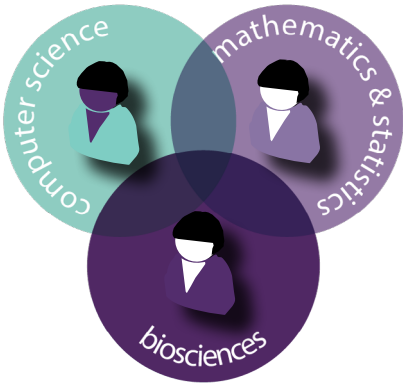
Author
Sobia Raza
sobia.raza@phgfoundation.org

Defining the role of a bioinformatician

Bioinformaticians are an increasingly important element of the healthcare workforce. As demand for their expertise grows, healthcare commissioners and providers need to consider how best to recruit, train, integrate, and manage bioinformaticians to ensure UK health services can realise the benefits of the 'big data' revolution.



The roots of bioinformatics for healthcare

The term 'bioinformatician' is often used flexibly to describe someone working in a broad range of clinical or research 'informatics' domains. Originally describing the study of informatics processes in biotic systems[1], bioinformatics today is a broad, interdisciplinary field that integrates principles from computer science, mathematics and statistics in order to manage, mine, visualize and analyse biological data. Bioinformatics has effectively co-evolved with disciplines such as genomics, and is now viewed as an integral component of these fields in academia.

The number of healthcare organisations facing big data challenges, for example analysing genome sequence data, is also increasing rapidly. The consequent escalation in demand for the skills of bioinformaticians has not, however, always been matched by an increase in understanding of the capabilities and limitations of bioinformatics, or by preparedness for integration of this new class of healthcare scientists into the workforce.

What then are the varied roles of bioinformaticians and what do UK health services need to do to maximise their skills for better, more effective patient care?

What are the key practical and policy considerations for UK organisations tasked with the delivery of health services and seeking to draw on the skills of bioinformaticians?

The objectives of bioinformatics

Bioinformatics is a broad discipline and often considered complementary, (if not equivalent) to the fields of computational biology and biostatistics. The overarching objectives of bioinformatics can broadly be categorised into the three key areas:

- analytical method development
- construction and curation of computational tools and databases
- data mining, interpretation and analysis

1. Analytical method development

The construction and refinement of mathematical algorithms and statistical methods for the management and analysis of biological and biomedical data.

Bioinformaticians function in a 'research and development' role, frequently applying statistical programming languages, mathematical modelling and computer simulations to design and implement methods for analysing data. *e.g.* the development of algorithms to detect mutations in our genomes.

2. Construction and curation of computational tools and databases

The collation, organisation and annotation of biological and medical data to aid its retrieval and analysis. Additionally the development and refinement of software tools that implement algorithms for analysing biological and biomedical data.

Bioinformaticians work in an 'engineering' capacity, typically using programming experience to develop bioinformatics workflows, databases and tools. *e.g.* the creation and maintenance of database resources cataloguing information on genes and proteins.

3. Data mining, interpretation and analysis

The extraction and analysis of data from databases or datasets using computational tools to derive biological or medical knowledge and insight from them.

Bioinformaticians apply the above computational tools and analytical methods, as well use computer programming languages to extrapolate biological 'meaning' from data. *e.g.* the application of bioinformatics tools to predict protein structure and function.

For a bioinformatician to deliver all these objectives would require a wide array of skills and deep understanding of many disparate domains of science. Like most scientists, individual bioinformaticians rarely possess the proficiencies to fulfil all the possible objectives of their field. Instead they specialise according to their specific expertise and education, and work with other bioinformaticians, scientists and health professionals to achieve the broader objectives of their work.

Recruiting and training bioinformaticians for health

At present individuals tend to enter the field from a range of undergraduate backgrounds (most commonly, biosciences, mathematics or computer science) by either learning appropriate skills and knowledge 'on the job' or undertaking postgraduate training. The multidisciplinary nature of bioinformatics requires practitioners to be flexible to learning new skills and acquiring new knowledge (*e.g.* programming languages or biology) as the scope of the data and analyses with which they are working evolve.

Definitions

Bioinformatics

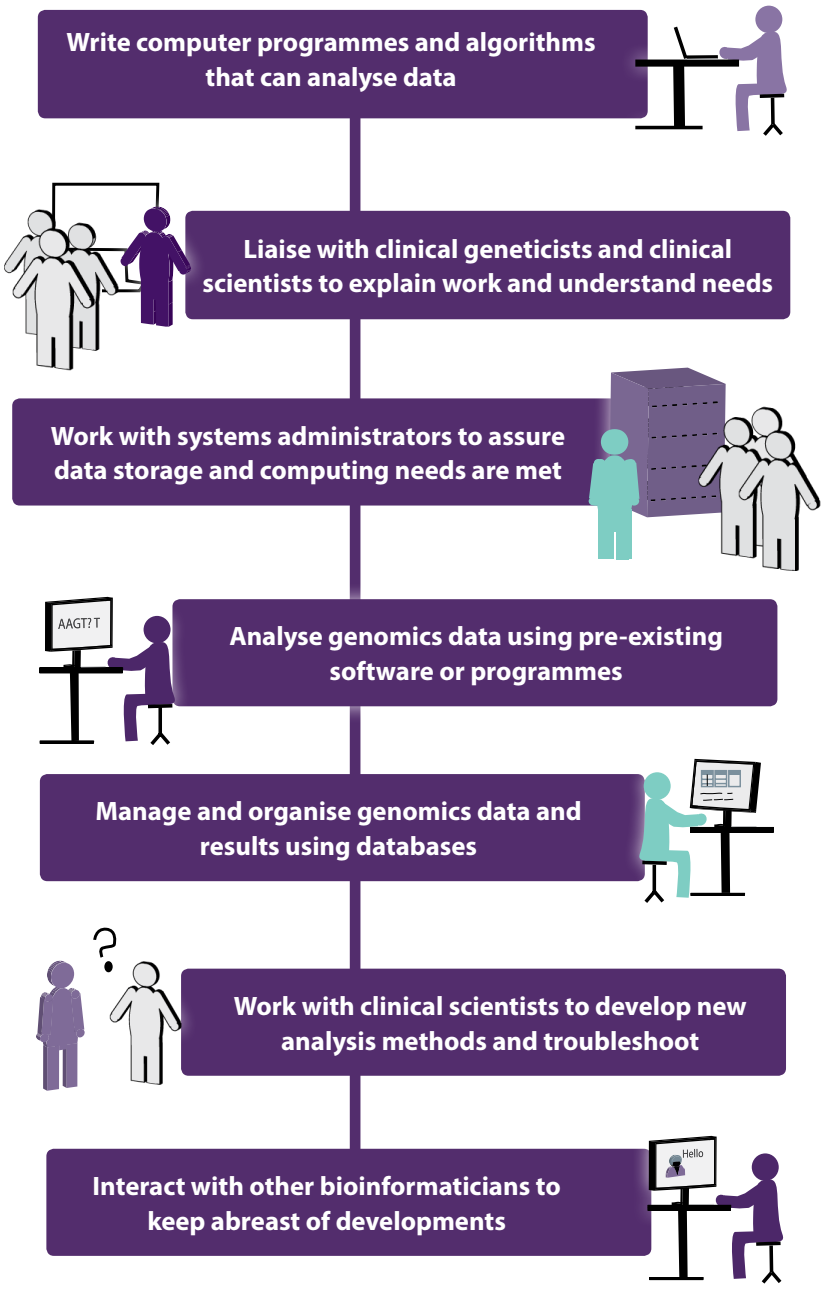
An interdisciplinary field which combines concepts and knowledge from computer science, statistics and biosciences in order to manage, mine, visualise and analyse biological and medical data.

Clinical bioinformatics

The clinical application of bioinformatics-associated sciences and tools to inform the medical management of human disease[2].

It is less common for bioinformaticians joining the healthcare sphere to have experience of clinical genetics and operation of the healthcare system. The recent NHS scientific training programme (STP) in bioinformatics and clinical bioinformatics is designed to be a training route for bioinformaticians within clinical specialities.

How bioinformaticians can support DNA sequence analysis in a clinical genetics laboratory



Definitions

Translational bioinformatics

The development and application of bioinformatics techniques to optimise the transformation of data (basic molecular, genetic, cellular, and clinical) into clinical products or health implications.[3,4]

Health (medical) informatics

Specifically, the application of informatics knowledge and tools to enable the collection and management of data for the delivery of health services.

A professional body?

As bioinformatic input has increasing impact on health service delivery, now is the time for a review of the standards of competence, ethics, conduct, and training needed for bioinformaticians in the health services.

Currently there is no recognised professional group for bioinformaticians, nor a requirement for their professional registration.

Consideration should be given to the desirability of professional registration and the establishment of a specialist group to determine standards of competency, ethics, conduct and training to ensure quality of services.

Actions for policy makers and employers

Given the broad remit of bioinformatics it is imperative for employers to recruit for their particular needs and assess how best to integrate bioinformaticians into the workplace. They need to ask themselves:

- **What is the task at hand?**

  Although generally bioinformaticians are proficient at computational assignments, their role can become confused with other distinct yet complementary fields. For example building a software tool with a user interface as part of a new diagnostic test (software engineer) or creating a website for accessing a repository of pathology test results (website developer) do not necessarily require a grounding in 'bio-sciences', and may be better suited to specifically trained individuals than bioinformaticians.

- **What 'type' of bioinformatician is needed?**

  Bioinformatician is a catch all term and may not be a useful descriptor when recruiting for different roles within health services - a 'bioinformatician' competent at creating databases and data archiving won't necessarily have the skills to analyse and interpret the datasets they create.

- **How many bioinformaticians?**

  It might be tempting to view bioinformaticians as a 'swiss army knife' profession where one 'tool' can perform a multiplicity of jobs. In reality, introduction of innovative analytical technologies, such as genomics and proteomics into healthcare are likely to require multiple bioinformaticians with different skill-sets at different stages of implementation and service delivery.

- **What equipment will be needed?**

  As with other scientific disciplines bioinformaticians require dedicated tools to perform their role. Access to suitable computational hardware, data storage capacity and servers, are just some of the fundamental structures.

- **How can we enable collaboration?**

  Fostering interaction between bioinformaticians in different organisations, both within the health service and in academia, will enable adoption of best practices and collaboration to deliver innovation. Promoting this may be particularly important where bioinformaticians are recruited in isolation.

- **How can we support continued professional development (CPD)?**

  Given the ever evolving nature of biosciences data, an environment conducive to CPD is key to attracting and retaining talent, as well as sustaining innovation.

References

1. Hogeweg P. PLoS Comput Biol 7, e1002021 (2011). 2. Wang X & Liotta LJ Clin Bioinforma 1, 1 (2011). 3. Kim JH. BMC Med Genomics 6 Suppl 2, I1 (2013). 4. Butte, AJ. J Am Med Inform Assoc 15, 709-14 (2008).