

*Presidency of the Council of Ministers*



## Abstract

### **MANAGING “INCIDENTAL FINDINGS” IN GENOMIC INVESTIGATIONS WITH NEW TECHNOLOGY PLATFORMS**

17 March 2016

In this new document on human genetics the NBC deals with the bioethical issues raised by the rapid evolution of second generation genome sequencing techniques, which over the recent years have transformed and speeded up research on and the diagnosis of many illnesses. These new techniques have posed the need for new shared guidelines and, in particular, standardised regulations for managing the so-called incidental findings, both at clinical and research levels.

Following a description of the state of the art, the NBC goes over a number of crucial ethical steps of the debate with particular reference to the discussion on the theoretical foundations of the 'right not to know' in a genetic context.

Even though aware that this transition from basic knowledge to clinical applications is characterised by a high degree of uncertainty and by knowledge that runs the risk of getting outdated in a short space of time, the NBC highlights some of the requirements for the genetics centres and the laboratories carrying out such tests. It stresses that the uncertain demarcation of the line between research and its clinical applications must never let one lose sight of the fact that the diagnostic tests are primarily aimed at giving a diagnosis to the patient, whose needs must remain at the centre of the investigation, and thus recommends that the patient/sample are included in a research project only when the diagnostic investigation has been concluded (either positively or negatively).

Furthermore, it recommends that the traditional distinction is made between adults and minors in clinical investigation and research and that the 'best interest' of the subject, not yet able to give their own consent, is placed under a particularly careful assessment. It is also recommended that, upon coming of age, the minor is contacted and able to choose to give/not give their consent to the further conservation of their samples and data (as already highlighted in the Opinion *Paediatric biobanks*).

As far as concerns the question of the returning of information to the donors of biological samples for research purposes, the NBC considers that, in the case of research foreseeing the gathering of large samples, it is unrealistic to contact the donors to update them on the results, which to date would hardly have any clinical significance of single interest. On the other hand, it considers that if requested it is morally dutiful to guarantee the return of the results of clinical relevance (incidental findings included) to patients with rare diseases and still without a certain diagnosis, always leaving the possibility of opting only to know some types of information to the individual.

Lastly the NBC stresses that the central role played by genetics and genomics in the healthcare panorama makes it increasingly urgent to rethink the training of the professional figures committed to this field of medicine and, at the same time, the organisation (starting from the school) of initiatives at various institutional levels aimed at citizens, in order to foster the acquisition of the necessary knowledge, which equally includes bioethical knowledge, so as to actively and critically tackle these transformations.

The Working group was coordinated by Profs. Monica Toraldo di Francia and Bruno Dallapiccola, who prepared the draft of the document.

The following took part in the drafting of the document and in particular Profs.: Silvio Garattini, Carlo Petrini and, in the working group discussion, Salvatore Amato, Carlo Casonato, Rosaria Conte, Lorenzo d'Avack, Antonio Da Re, Demetrio Neri, Laura Palazzani, Grazia Zuffa.

The group met for the first time on 19 June 2014 and, after various meetings, the Opinion was debated in the plenary session on 26 February 2016.

The expert advice of Prof. Dallapiccola was given on "The point on personalised medicine (genomic analyses of complex illnesses)" and by Prof. Alberto Piazza.

The document was approved by the members present, Profs.: Amato, Battaglia, Canestrari, Caporale, d'Avack, Da Re, De Curtis, Di Segni, Flamigni, Frati, Gensabella, Morresi, Neri, Nicolussi, Sargiacomo, Scaraffia, Toraldo Di Francia, Zuffa.

The members without the right to vote expressed themselves in favour Drs.: Benato, Bernasconi, Conte, Petrini.

Profs. Caltagirone, Casonato, D'Agostino, Dallapiccola, Garattini, Palamara, Palazzani, Proietti endorsed the Opinion at a later date.