

*Presidenza del Consiglio dei Ministri*



**ORPHAN DRUGS FOR PERSONS AFFECTED BY RARE  
DISEASES**

25<sup>th</sup> of November 2011

**Abstract**

The document analyses the difficulties faced by people affected by rare diseases which still pose a challenge as regards diagnosis, the limitations of aid and therapy, which, in most cases, is non-existent. The opinion focuses on the statistical data in reference to the rare diseases currently present in Italy and internationally and the orphan drugs available in this context. The NBC emphasizes how the rarity of the disease greatly reduces investment by pharmaceutical industries, owing to the scarcity of economic returns. Therefore, the problem can not be tackled on a national level, but must assume a European and international dimension.

The NBC, while recognizing the difficult solution of the problem, proposes some measures in order to limit it and ensure - as far as possible - the conditions of justice; promotion and economic support of research, by public and private structures, aimed at achieving a better knowledge of rare diseases, ensuring the quality of drugs and the creation of new ones; the careful control of expenditure for these kinds of drugs, which, as a result of their being directed at neglected areas, are entitled to concessions and benefits, and therefore can give rise to a waste of resources or speculation; greater coordination in the search for genetic abnormalities with the appropriate development of genetic counseling and genetic therapies; the reduction of the threshold that defines the rarity of disease to ensure the sustainable promotion of research, development, marketing and delivery of truly innovative drugs.

The NBC also recommends monitoring the effectiveness and tolerability of drugs granted for compassionate use or used in an *off-label* form; the recovery of resources in order to sustain the burden of orphan treatments through the redistribution of expenditure for some classes of widely used and low cost drugs, from the NHS to patients; and the promotion of campaigns in order to encourage large companies to 'adopt' one of the orphan diseases.