

AISAC - Association for Achondroplasia Information and Study

In reply to the Public Consultation, AISAC would like to highlight:

The strategic, political and social importance of the involvement of patient associations, with “lay” administration (not subordinate to doctors), whose experience in caring for patients and widespread presence on the ground throughout Europe can ensure constantly up-to-date and efficient transmission of knowledge and information.

The importance of ensuring that European citizens suffering from rare diseases have equal rights and opportunities in the medical/scientific, rehabilitative and social fields, so that they are not discriminated against.

The importance of a commitment on the part of the reference networks of centres of expertise to simplifying information management so as to create, for example, a single parlance/common code which is accessible and comprehensible in all States.

The importance of providing individuals with a holistic care approach and, accordingly, for the Member States to pay increased attention to socialisation, schooling, social inclusion and rehabilitation, so as to improve the quality of everyday life of people suffering from rare diseases.

The importance of simplifying bureaucratic, organisational and political processes so as to prevent them from becoming an obstacle to the effectiveness and efficiency of the activities carried out.

The importance of common guidelines across Europe for the management of care protocols, so as to ensure that best practices become shared assets.

More specifically, AISAC’s replies to your questions are as follows:

**Question 1:**

We consider the definition satisfactory.

**Question 2:**

We agree that it is necessary to improve and standardise, at European level, coding and classification of rare diseases and we hope that, in all such decisions, precedence will be given to criteria of conceptual and operational simplification.

**Question 3:**

We agree, particularly in relation to the “rarest” diseases.

**Question 4/5:**

As regards the dissemination of correct information, we consider it absolutely pointless to duplicate work, and therefore we are in favour of the financial agreement between Orphanet and the EU.

We also agree with the programme mentioned concerning support to the information networks and the development of national/regional reference centres, so as to avoid patients being moved, and EU reference networks, so as to ensure equal treatment of individual diseases throughout the EU. However, this requires a firm political commitment from all Member States.

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In our opinion, the problem requires equal dissemination of knowledge throughout Europe, including through the information networks. The European reference networks should give priority to the transfer of knowledge, including through research to further develop distance diagnosis. Universities should play an active role in raising awareness on issues linked to rare diseases (initial notification of diagnosis, choice of care approach, guidelines, etc.).

We believe it is extremely important to avoid patient movements by facilitating the adoption and use by all parties of online and electronic instruments that help increase the chances of good care provision in patients' place of residence.

We agree that the adoption by the EU of all forms of online and electronic tools should be considered a strong point of the strategy.

**Question 6:**

AISAC believes that it is very important to pursue policies encouraging European-level supervision laboratory quality.

**Question 7:**

We believe that screening should be validated at European level.

**Question 8:**

With regard to the application of primary prevention measures, the implementation of best practices in healthcare and equality of access to orphan drugs, AISAC is in full agreement, but wishes to stress that, as regards access to drugs, there need to be political decisions at national level based on the right of each individual to receive the best care possible even if, or precisely because, he/she suffers from a rare disease.

**Question 9:**

AISAC agrees that the EU should have a regulation concerning medical devices and diagnostics.

**Question 10:**

The reply to this question should be found within a careful examination concerning the existence and availability of specialised support services in each Member State for anyone suffering from genetically complex diseases which cause disabilities and serious impediments to leading a normal life. Therefore, we are in favour of funding to strengthen and improve the quality of the existing services for all citizens and against the creation of additional specific facilities.

**Question 14:**

AISAC considers it very important to support patient associations and the implementation of a fair policy towards meeting the needs of people suffering from rare diseases. For example, this year in Italy, the Minister for Health, Livia Turco, established the "National Council for Rare Diseases" at the Ministry of Health. This body is made up of 34 associations from various Italian regions and is coordinated by Dr Taruscio from the National Institute of Health. These associations, which have been working together for some time on further research into the current problems in Italy concerning clinical and regulatory care for people suffering from rare diseases and possible solutions to these problems, submitted their report to the Minister, who took it into account as it had been written by "a group of people considered to be the *main experts* in the specific diseases".

On behalf of AISAC: Donatella and Marco Sessa

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