



Title Description of the Current Status of Prenatal Screening in the Most Frequent Fetal Chromosopathies, Mainly Down Syndrome in Spain, and Proposals for the Improvement of Everyday Clinical Practice

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Aim

To describe the current status of screening and prenatal diagnosis of Down syndrome (DS) in autonomous communities in Spain; to assess the effectiveness and safety of different approaches to prenatal screening for DS by ultrasonography and serum markers; and to do a cost-effectiveness analysis of the implementation of remedial screening for DS.

Methods

Spain was considered the geographical framework in calculating the following: a) Overall cost of the program for a sample of 100 000 pregnant women and a DS prevalence of 25/10 000 gestations (EUROCAT, 2006) and b) Cost per case of confirmed DS.

Please consult the report for further information.

Conclusions and results

1. Most autonomous Spanish communities do not have a clear strategy for screening and prenatal diagnosis. Written information sources are deficient, and the data obtained via telephone did not improve the quality. Hence, it is urgent to establish/update programs to screen and diagnose the most common fetal chromosomal disorders, particularly DS, in autonomous communities where programs do not exist, or are obsolete. The available scientific evidence should be considered, particularly the opinions of Spanish experts and patients, when establishing the objectives of this National Health System project.
2. Methods that include markers of the first and second trimesters are most effective and safe. Nevertheless, to decide on a prenatal screening policy to implement, we must assess not only effectiveness and safety, but also cost effectiveness and women's preferences and values.
3. The *contingent sequential test* offers the best cost-effectiveness ratio and intermediate diagnostic performance. Of the alternatives analyzed, the *combined test* involves a higher overall cost of imple-

mentation and is the least cost-effective, with a cost of 5 617 361 euros (EUR) and EUR 27 740 per confirmed DS, respectively.

4. Drawing up a series of minimum recommendations and coordinating the agencies, scientific societies, and specialists involved in the process was deemed to be fundamentally important to ensure implementation of a quality control program for screening throughout Spain.
5. Nearly all Spanish women agree with implementing a national program of prenatal screening for chromosomopathies. They stress the importance of assessing the reduction of unnecessary invasive procedures. The personal recommendation of physicians is another important element in the decision-making process by pregnant women.