

The American Society of Human Genetics

Policy Statement

ASHG's Response to NIH Office of Science Policy on Draft Guidelines for Research Involving Human Pluripotent Stem Cells

January 21, 2000

NIH Office of Science Policy
1 Center Drive Bldg 1, Rm 218
Bethesda, MD 20892

RE: Draft NIH Guidelines for Research Involving Human Pluripotent Stem Cells

To whom it may concern:

I write on behalf of the [American Society of Human Genetics \(ASHG\)](#), which represents about 7,000 biomedical researchers across the country and throughout the world, regarding the December 2, 1999, "Draft National Institutes of Health Guidelines for Research Involving Human Pluripotent Stem Cells."

The guidelines are an important first step forward toward enabling federally-funded scientists to conduct research on human stem cell lines, and thus to provide the maximum medical and scientific benefit to the American people. We especially commend the thoughtful balance reflected in the guidelines between issues of science and issues of ethics.

The identification of embryonic stem cells is a major scientific achievement with enormous potential related to the treatment of human disease. Realization of the potential, however, requires ongoing research, and the proposed guidelines will enable this critical research to advance without compromising the moral and ethical values of the majority of Americans. Therefore we support the guidelines in general.

In particular we would like to register our support for three critical points.

1. The ASHG supports the proposal that NIH funds can be used for research on pluripotent stem cells only if they have been derived from early human embryos that were created for the purpose of infertility treatment and were in excess of clinical need.
2. The ASHG supports the requirement that the donation of human embryos be voluntary and not recompensed at the time of donation or after stem cell lines have been generated from them.
3. The ASHG supports the formation of the Human Pluripotent Stem Cell Review Group (HPSCRG) to oversee the guidelines.

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As researchers in the field of human genetics, members of our Society are active in research designed to relieve human suffering caused by genetic alterations. While great advances in understanding genetic disease have been made in the last two decades, aided by the Human Genome Project in the last decade, there are still many common diseases whose genetic alterations remain to be determined, and for those whose defects are known, there is still a need for research to find desperately needed treatments or cures.

To these ends, researchers in human genetics often use cells or cell lines derived from individuals who are genetically or chromosomally abnormal. Stem cell lines derived from such abnormal embryos will be extremely valuable to study disease mechanisms. There are two potential sources of abnormal embryos. First, in the course of IVF treatment, embryos are obtained that are not suitable for implantation. These are either identified microscopically as structurally abnormal or nonviable and/or by fluorescence in situ hybridization as chromosomally imbalanced (having chromosomal defects). Such embryos are usually discarded and not frozen. Second, preimplantation genetic diagnosis (PGD) is performed for couples with a known genetic risk for abnormal embryos who do not wish to undergo prenatal diagnosis later in pregnancy. Blastomeres obtained from IVF-generated embryos of such couples are tested for the presence of the genetic defect and only those that are unaffected are implanted or frozen, while those affected are usually discarded. There is no medical reason for freezing the abnormal embryos. Their chance of recovery from freezer storage may also be reduced because of their genetic or chromosomal defect.

We therefore suggest two critical changes to the draft guidelines that would allow research on genetically defective embryonic stem cells without, we believe, further compromising the ethical principles upon which the guidelines were developed. These suggestions are as follows:

1. Allow the use of genetically or chromosomally abnormal embryos for the derivation of pluripotent cell lines without the requirement that they first be frozen. The procedures and protocols for the use of such abnormal fresh embryos for the establishment of ES cells will have to be clearly specified.
2. Alter the requirement that "all identifiers be removed" to allow the genetic or chromosomal diagnosis (including identity of mutation, if known) to be retained. Identifying a genetic or chromosomal mutation does not identify one particular individual or family.
3. We appreciate your development of these guidelines and the opportunity to comment on them. Amendment of the draft guidelines, as recommended here, will enable the use of unique, valuable tissue resources for human genetic research. The discovery of disease genes and mechanisms is essential for the development of novel diagnostic and treatment modalities.

Finally, we have some general concerns on the practicality of some of the guidelines as pointed out by the American Society for Cell Biology (ASCB) in their response to the guidelines. We agree with the ASCB that the guidelines need to address the difficulty of validating the

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procedures and conditions used in creating the cell lines, and that once a cell line is validated it should not have to be revalidated by each user.

Sincerely,

Uta Francke, MD
Past President, ASHG

Ronald Worton, PhD
President, ASHG