MAKATI MEDICAL CENTER
BIOETHICS EDUCATIONAL COMMITTEE

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CASE STUDY 33

GENETIC COUNSELING

CASE

A baby girl was born 6 months ago via normal spontaneous delivery, full term, to a 32-year-old G4P3 mother, with no fetomaternal complications. At age 3 months she showed no head control, unable to sit even with support, and with generalized hypotonia. At six months she was hooked to a mechanical ventilator, and all attempts at weaning failed. Muscle biopsy revealed fascicles of atrophic Type I and Type II muscle fibers, interspersed with small clusters of hypertrophic of Type I muscle fibers consistent with Werdnig-Hoffman's disease.

The attending doctor explained to the parents that this incurable disease progresses chronically, makes weaning the patient from the ventilator impossible, and usually leads to death within 6-24 months. Aside from discussing the present case, the doctor also recommended genetic counseling to the parents.

QUESTION

What is genetic counseling?

POSSIBLE SOLUTION

At the present stage of medical science, many genetically transmitted diseases can be predicted, or at least their probable incidence could be foreseen. This occurs by analyzing the parents' genetic make-up and/or observing disease patterns of ascendants, other children and relatives.

Genetic counseling offers parents information on probabilities of genetic defects in their children, the consequences of these defects, and the therapy and care that follows. Genetic counselors also help parents, whether directly or by referral to others who possess professional competence, and assess the available personal, economic and social factors and resources.

Parents, of course, make the final decision whether to have children or not, taking into account:

1. Their felt desire and need to have children.
2. Risks of genetic defects.
3. Their capacity to take care of these children.
4. The burden to the children themselves suffering from severe handicaps.
5. The risk that these children would in turn pass on defective genes.

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IDEA ETHICAL DECISION-MAKING FRAMEWORK & WORKSHEET

SCAN HERE

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