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SPECIAL ARTICLE

The Impacts of Genetic Counseling on The Quality of Children

by

SJARIF HIDAJAT EFFENDI

(From the Department of pediatrics, Padjajaran University, Bandung)

Introduction

The success of our National Family Planning Program and awareness of people to the motto of: "The norm of a small, happy and prosperous family" has led our people, especially the eligible couples, to think no more about quantity, but the quality of their children. There is a tendency that they do need assurance of completely healty children, the present or the coming ones. This fact leads our Health Service Personnel to the awareness that they must be able to solve people's problems related to their desire for good quality children.

Increasing the children's quality of life greatly depends on the efforts within the following stages of their lives: conception, pregnancy, delivery and neonatal conditions. It means that marriage/genetic counseling, antenatal care, and perinatal services play a very important role. Every disturbance within these periods will lead to serious damage to the growth and development of organs, especially the brain. This condition might result in handicapped and malformed children.

As infectious disease are brought under control and decreasing, the relative importance of overcoming disorders that are wholly or partly genetic has been a decrease in the perinatal.Mortality rate, incidence of infections and malnutrition in hospitals in Indonesia, but cases of congenital hereditary disorders are increase.

Every year thousands of familiels are effected by the birth of an abnormal child. About 0.5 % of newborns have chromosom abnormality with moderate to severe phenotypic effects.


Received : October 14, 1991
Another 0.5 - 1% suffer from the consequences of single-gene defects (dominant, recessive, or x-linked), and about 2% have malformation that may in part be due to inheritable factors. Altogether, about 4% of new borns have a serious defect that is recognized at birth or within their first year. It has been estimated that about a third of all children in pediatric hospitals are being treated for conditions that have genetic component. Thus there can be no doubt about the need for increased genetic information and counseling many families requiring it.

In countries with low infant mortality rates the cause of death are mostly perinatal problems and congenital disorders. That is why the high incidence of the disease during the perinatal period and the high neonatal and perinatal death rates make it important to identify the fetus and infant risk as early as possible.

Causes of congenital malformation are genetic and non-genetic factors. The non-genetic factors are such as uterine abnormalities, teratogens, and poor maternal conditions.

This paper discusses some aspects of genetic counseling including its process, organization, goal, and its role in promoting the quality of children.

### Congenital Genetic Abnormality

About 3% children in developed countries are mentally retarded. The ratio of male and female is about 1.5-2 : 1. About one fifth of those mentally retarded children have a chromosomal basis for developmental retardations.

A useful classification of disesase having a genetic background would be:

1. Single mutant gene (mendelian) disorders
2. Chromosomal aberration syndromes
3. Diseases determined by multifactorial inheritance-genetic predisposition with environmental interaction
4. Somatic cell genetic diseases (neoplasia, aging, autoimmune disease, some congenital malformations)

At this time there are about 4300 diseases having a genetic basis. It is increasing dramatically i.e.: 1500 in 1986, 2800 in 1978, 3300, in 1983, and 3900 in 1986.

### Genetic Counseling

Genetic counseling is the process by which patients or relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorders, the probability of developing and transmitting it among the ways in which this may be prevented or ameliorated.

Based on this definition there are 3 aspects of genetic counseling:

1. The diagnostic aspect without which all advice will be baseless and presumptuous. There is no alternative to exact diagnosis. To assess genetic risk one needs a precise, accurate, and detailed family history apart from the diagnosis;
2. The actual estimation of risks which may be simple in some situations and complex in others;
3. A supportive role, ensuring that the consultee actually benefits from the advice given and from the various preventive measures that may be available.

Vitamin bayi lengkap, membantu bayi tumbuh kembang, sehat, kuat dan cerdas

8 vitamin esensial dalam Abdec Drops membantu pertumbuhan optimum bayi. Abdec Drops dalam bentuk cairan, bebas alkoalol serta rasa apel lezat, sangat disukai bayi. Cara pemberianinya mudah, dengan dosis sekali sehari – langsung atau dicampur minuman makanan bayi.

<table>
<thead>
<tr>
<th>Komposisi</th>
<th>Dosis</th>
<th>Kemasan</th>
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<tbody>
<tr>
<td>Tiap 0,6 ml mengandung:</td>
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<tr>
<td>Vitamin A</td>
<td>5000 unit</td>
<td>Bayi (kurang dari 1 tahun): 0,3 ml per hari, 0,6 ml per hari.</td>
</tr>
<tr>
<td>Vitamin D</td>
<td>440 unit</td>
<td>Anak-anak (lebih dari 1 tahun): 0,6 ml per hari.</td>
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<tr>
<td>Vitamin B1</td>
<td>1 mg</td>
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<tr>
<td>Vitamin B2</td>
<td>1,2 mg</td>
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<tr>
<td>Vitamin B6</td>
<td>1 mg</td>
<td></td>
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<tr>
<td>Nikotinamida</td>
<td>10 mg</td>
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<tr>
<td>Asam Pantothenat</td>
<td>5 mg</td>
<td></td>
</tr>
<tr>
<td>Vitamin C</td>
<td>50 mg</td>
<td>Botol bensin 15 ml.</td>
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* Merek dagang terdaftar.
The aim of genetic counseling is to convey medical and genetic facts to the affected or potentially affected family and to explain the options available. However, it involves much more than simply reeling off statements of probability.

Figure 1. Depicts the stages of genetic counseling process.

It begins with a questions raised by the advent of a child with the possibility of a genetic disorder, other aspects of the family history, etc. There must be a diagnosis which may depend on information from clinicians, syndromologist, X-ray studies, laboratory test, dermatoglyphics, cytogenetics and the family history. To answer the question usually requires estimating probability using information from the family history, cytogenetics, the literature, the mendelian principles and Bayesia calculations. Following informative and supportive counseling (often influenced by social, moral, economic, and family pressure) the counselee may reach a decision either to refrain from reproduction or to go ahead. Either of these may require appropriate referral. If the "Go" decision results in a recurrence, further counseling may be required. Follow-up of the counselee and the extended family may result in reentry into the process.

From the facts mentioned above, it appears that the corner-stone of genetic counseling is a correct diagnosis. This
precise and accurate diagnosis can only be obtained by establishing a good, professional and multidisciplinary cooperation among the pediatricians, obstetricians, syndromologists, molecular biologists, biochemists, cytogenetics, radiologists, neurologists, cardiologists, social workers, psychiatrists, and so on.

**Organization**

The activities of genetic counseling should be well organized together with other disciplines supporting Clinical Genetics as in the following example:

A fact is that it is not possible to set up a genetic clinic as an individual, as cooperation of multidisciplinary examinations is an absolute requirement.

**Figure 2. The Organization of Clinical Genetics**

<table>
<thead>
<tr>
<th>Clinical Genetics Department</th>
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<tbody>
<tr>
<td>Director</td>
</tr>
<tr>
<td>Administration --------------</td>
</tr>
<tr>
<td>Cell bank</td>
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<tr>
<td>Computer</td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Metabolic disorder</th>
<th>Metabolic lab</th>
<th>Enzymology lab</th>
<th>DNA Biochemical lab</th>
<th>Chromosome lab</th>
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| Department of pediatrics    | Department of Cellbiology and Genetics | Genetic Counseling of Ob.Gyn |

**Prenatal Diagnosis**

The goal of genetic counseling in relation to ensure the quality of children is the action of prevention, whether it is done by prenatal diagnostics or calculating the recurrence risk.

It is becoming increasingly possible to evaluate the developing fetus in an attempt to detect serious disorders early enough to allow for termination of affected pregnancies when there is significant risk, often a recurrence risk of serious disorder.

5. Genetic risk to pregnancy sufficiently high

**The Role of Genetic Counseling in Promoting the Quality of Children**

The impact of a diagnosis on the consultee can cause a variety of reactions. They include anger, guilt, shame and blame casting. The first stage of shock and disorganization is soon taken over by the second stage of rationalization, denial and blame casting. Finally, when they reach the third stage of equalization, they are able to act more realistic and function more effectively.

One's perception of risk and willingness to accept it are highly subjective matters, depending on one's personality, experiences, moral convictions, and especially on the burden of care imposed by the condition in question.

When the genetic prognosis is unfavorable, a couple may consider only two options for further action: to refrain from childbearing or to risk a defective birth. If aborting is an acceptable alternative, however, prenatal diagnosis followed by possible termination of pregnancy is now available in a growing number of cases. Alternatively, a couple may choose adoption or an appropriate reproductive technology using either a donor egg or donor sperm.

When this procedure of genetic counseling is completely done, it means that we can as maximal as possible eliminate the birth of congenital genetic malformed neonates.

**Conclusions and Suggestions**

As perinatal problems are brought under control, the relative importance of overcoming genetic disorders has been steadily increasing.

Genetic counseling (a tool to solve those genetic problems) can ensure to eliminate or prevent genetic disorders, supporting good quality children to mankind. But to set up a genetic counseling activity a good multidisciplinary cooperation and collaboration is needed.

The activity of genetic counseling, however, needs and accurate diagnosis as a cornerstone.

To face these coming problems, education of young basic scientists should be started from now on. Especially in Indonesia, the question arises whether the curriculum of the medical Faculty which is at present for a great part based on community oriented medicine, should be changed gradually towards a more basic science program such as biochemistry, cytobiology, genetics, etc.
REFERENCES


SELECTED ABSTRACTS


The clinical characteristics of acute otitis media in relation to coexisting respiratory virus infection were studied in a 1-year prospective study of 363 children with acute otitis media. Respiratory viruses were detected using virus isolation and virus antigen detection in nasopharyngeal specimens of 42% of the patients at the time of diagnosis. Rhinovirus (24%) and respiratory syncytial virus (13%) were the two most common viruses detected. Adenovirus, parainfluenza viruses, and coronavirus OC43 were found less frequently. The mean duration of preceding symptoms was 5.9 days before the diagnosis of acute otitis media. Ninety-four percent of the children had symptoms of upper respiratory tract infection. Fever was reported in 55% and earache in 47% of cases. Patients with respiratory syncytial virus infection had fever, cough, and vomiting significantly more often than patients with rhinovirus infection or virus negative patients. No significant differences were found in the appearance of the tympanic membrane and the onset of illness between virus-negative and virus-positive patients with acute otitis. Most patients respond well to antimicrobial therapy despite the coexisting viral infection. If the symptoms of infection persist, they can be due to the underlying viral infection, and viral diagnostics preferably with rapid methods may be clinically useful in these patients.


Homeless children in families are increasing in number across the country and have been noted to have frequent health problems. The health status of homeless children was assessed on multiple dimensions through parental report in a survey conducted with 196 homeless families in 10 shelters in Los Angeles and 194 housed poor families after March 1987 through January 1988. During the month before the survey, the homeless and housed poor children experienced high rates of illness symptoms, disability, and bed days. Homeless and housed poor children were frequently rated by their parents to be in fair or poor health (17% vs 13%, $P = 14$). Homeless children, however, were reported to have more behavior problems and school failure (30% vs 18%, $P = 06$) than housed poor children. Homeless children also had high rates of other health problems such as developmental delay (5%) and overweight (13%). The diets of homeless children were frequently imbalanced, dependent on food from fast-food restaurants, and characterized by repeated periods of deprivation. Family problems were more common among homeless families, especially among single-parent homeless families compared with single-parent housed families (spousal abuse, 68% vs 41%, $P < 01$; parental drug and alcohol abuse, 60% vs 39%, $P < 01$). It is concluded that homeless children have significant child behavior and developmental problems and disorders of nutrition and growth, which are associated with multiple risk factors in their environment.