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Dear Beate

A letter about the non-sense of genetic counseling

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Dear Beate,

since many of your friends expecting a baby suddenly talk about “amniocentesis”, “high risk of a handicapped child” and stress that at least one should be well informed about these matters, you were confused and asked me what all this stuff is about.

As you know, I have studied human genetics and, out of my work and my experience, question the conviction that genetics can say anything meaningful for people like you and me. During my studies, I have seized the opportunity to listen to several genetic counseling sessions. As a critical and distant observer, one thing always struck me: Counselors, informing about birth risks and chromosomes, talked about something radically different from the stories and worries of the counselees. I started to wonder if this new kind of medical encounter in order to produce an “informed decision” could still be called “counseling”, with both sides talking at cross purposes. This is why I am reflecting, first on the differences between everyday experience and scientific discourse and, secondly, on the question what happens when both spheres clash. I want to find out how scientific terms are used to impose a certain kind of public health policy, and how the everyday experience of pregnant women like your friends, for example, is shaped by a professionally introduced medical-genetic way of thinking. Or, in a word: I want to know since when genetic risk carriers can dirty their diapers.

Now, in this letter, I will answer the question about prenatal testing and genetic counseling as to a friend who is at least sceptical about the promises of modern medicine and who did not yourself undergo amniocentesis and genetic counseling during your pregnancy. You wonder why many of your friends consider these things important and, suddenly, you are not “up to date” anymore. It is a very complicated question *why* so many women undergo all these examinations. Therefore, in this letter, I stay to your question *what* this stuff is all about.

In Germany, only physicians with a special training in the science of hereditary, medical statistics and molecular genetics can become genetic counselors. Genetic counseling is a new vocation, especially for women, because they are supposed to have better counseling skills such as empathy and sensitiveness. Generally caring about their clients` welfare, they speak with expertise to pregnant women about the uncertainties of a future birth. So, a genetic counseling session is about a textbook prognosis for a fetus at risk.

The more prenatal monitoring is intensified, the more pregnancies are supposed to be risky. Today, in Germany, gynecologists put 60-80% of all pregnant women into the class of “pregnancy at risk”. After your first medical check up when you were pregnant with your daughter Anna, you probably realized how quickly a woman can be impregnated with risk.
I still remember the beginning of your pregnancy very well: You experienced three months of doubt and nausea, and you felt very unsure of yourself with all the physical changes you lived through.Your belly grew, and you slowly became familiar with the idea of expecting a child.
In Germany, the official physician guidelines[1] call for an ultrasound between the 19th and 22nd week of pregnancy in order to detect any developmental disorders or malformation of the fetus. On the average, German gynecologists have a look at the woman’s belly with their ultrasound scanner six times during a pregnancy. So, these inspections have become a routine matter. Some physicians additionally run tests on blood samples in the 16. - 18. week of pregnancy. They measure the concentrations of three substances, including Alpha-Feto-Proteine, in order to detect potential (and numerous) deviations from norm. With a computer program, they calculate ratios like 1:935 or 1:110 which show the probability of the child being born with Down’s syndrome or spina bifida.

[1] The so-called “Mutterschaftsrichlinien”, revised version of 1996

This procedure, called “triple-test”, is a preselection of women for amniocentesis by means of statistics. Very often, women receive a bad test result saying the probability of the child having Down’s syndrome is very high, but in the end, the child turns out to be quite normal.

Just imagine your feeling: The gynecologist does not send you home with the reassuring remark, “everything’s OK”; rather, he wrinkles his forehead anxiously and suggests that something is wrong. The probability of a malformed child being born is higher than the calculated limit set by medical experts, or the physician cannot find the fifth finger on the ultrasound screen. In this situation, many physicians simply recommend or perform an amniocentesis without further ado. But your gynecologist is very much aware of modern bioethical values, and wants you to make an “autonomous decision”. In order to clarify what the implications of these probabilities are, he refers you to a genetic counseling center.

On a windy morning, you and your boy friend stride through the doors of the Institute of Human Genetics. In the spacious corridor, show cases exhibit skulls of different species of monkeys, and bone fragments of Homo neanderthalensis. These few exhibits refer to a time when anthropology was thought to reveal something about human genetics on the basis of appearances, the phenotype. This was before the double helix (DNA). Genetics as the science of hereditary phenomena came into existence at the beginning of the 20th century, in the context of social Darwinism and the growing eugenics movement. At that time, the crucial issue was to halt “degeneration” - as defined by geneticists and anthropologists - by educating the public through journals, lectures and counseling. In many countries, forced sterilization was practiced. A menacing picture of the decline of humankind because of hereditary factors was nurtured by abstract mathematical models of population, and by the results of research on fruit flies (drosophila). The eugenic vision was to increase the quality of the human gene pool by planned breeding in order to create better human beings.The means advocated by the eugenics movement was a socio-political regulation of human procreation on the basis of science.

Following the sign directing you to “Genetic Counseling”, you suddenly find yourself in the brightly lighted corridor of a modern 1990s laboratory. Men and women in white coats transfer test tubes from one room to another, conjure up a meaning out of a transparent liquid, and speak a for you incomprehensible language - lab jargon. You take a seat until a friendly middle-aged woman rushes up to you, quickly introduces herself, leads you out of the laboratory, past the monkey skulls, up a stair into a small room. The genetic counselor offers you and your friend a chair, and takes her seat on the opposite side of the desk. On the shelves there are dozens of books with “human genetics”, “birth defects” and similar words in their titles. Colorful pictures, apparently painted by children, hang on the walls.

The session starts with the counselor summarizing how all of you came to be here. The result of the Triple-Test estimates your probability to deliver a child with Down’s syndrome to be 1:110. This ratio is higher than the average frequency at your age; therefore, you are classified as a pregnancy at risk. The physician then clarifies that genetic counseling does not mean that she will advise you. You yourself have to decide the right thing for you to do. Today, genetic counseling is non-directive (which means, in practice, that the counselor is as much at a loss as you are). This concept, based on the idea of the autonomous individual, distinguishes present genetic counseling from its predecessors in the time of eugenics. The human geneticist Sheldon Reed[2] explicitly introduced the term, genetic counseling, in 1947, to dissociate himself from eugenics population policy, and to stress help for the individual. Reed counseled families actually or supposedly having a hereditary disease. He saw his work as “a kind of genetic social work” with no association to eugenics. According to Reed, the introduction of this new term made possible the establishement of the present genetic counseling institutions: “It is in my impression that my practice of divorcing the two concepts of eugenics and genetic counseling contributed to the rapid growth of genetic counseling.

[2] Reed, Sheldon (1974). A short history of genetic counseling. Social biology 21 (4): 332-339

Genetic counseling would have been rejected, in all probability, if it had been presented as technique of eugenics".[3]

Counseling today does not mean to give advice, but to "address possibilities"[4]. Whether it concerns the risk of a handicapped child, or the possibility of sperm donation in order to decrease the risk; whether it concerns the question of abortion in order to avoid any birth risk, or a further examination to obtain detailed diagnostic data - counseling only lays out the probabilities. In order to inform you, everything has to be mentioned and then you have to decide. Before you choose one of their options, the counselor will lay the basis, respecting your "autonomous decision".

The geneticist starts to question you about your relatives. This kind of medical interrogation is called pedigree analysis. All members of your family are recorded on a sheet of paper, including their age, their spouses, their offspring, important diseases and year and cause of their death. Your restless Uncle Paul, who never knew whether he was coming or going, becomes an abstract square on the paper, and his adventure filled and colorful life with your aunt a linear connection with a certain output: four more geometrical figures. She wants to know if everyone in your family is healthy, and if someone has died she asks for the reasons. In any strange or peculiar events, she searches for hints of a genetic burden. Every personal life story is transformed into a medical case history. Your grandmother, who usually became ill after sumptuous family feasts, and thereupon remained in bed for a day, becomes a diabetic. Diabetes has something to do with genes. Therefore, a correlation was made between all grandmothers with bouts of sickness and pregnant women's blood sugar level deviating from the average. Then, this abnormal blood sugar level is correlated with the frequency of a defined class of malformations. The result of such a statistical operation is understood as a risk. Then, since your grandmother belongs to the population of diabetic grandmothers, you are defined as being at risk, and your blood sugar level must be carefully monitored.

The daughter of your brother has hay fever. Your unborn child runs a 5 to 10% risk of getting hay fever, neurodermitis or asthma. Your aunt, like the mother of your father, died with a growth in her breast. Breast cancer can be at least partly hereditary, explains the concerned physician. You should regularly take advantage of preventive care to "minimize your risk".

Your boy friend comes from western Kenya, where children are simply born. He grew up with his nephews and cousins, as with his own sisters and brothers. Biological cause and effects did not have any meaning. Since his long absence from home, he only counted the male offspring of his sister, but not her daughters. His father and mother are distantly related, but details are wrapped in obscurity because of a tragic family story. When the geneticist asks your friend if he has fathered children with another woman, he only shrugs his shoulders with embarrassment. The counselor draws numerous squares and confused lines on the paper…and sighs.

Now you know how risky relatives can be. Every growth and allergy becomes a risk for you and your offspring. Yet, to ascribe a risk to somebody is approximately as meaningful as the German saying: "Wenn der Hahn kräht auf dem Mist, ändert sich's Wetter, oder's bleibt, wie's ist" (when the cock crows on the manure pile, the weather changes or stays the same). For you, a risk means nothing else but: it may happen or not. Ten percent breast cancer does not exist. But suddenly possibilities strike you - fears heretofore completely foreign to you: Is the fate to live and die with a growth like my aunt's programmed in my body? When my child is born, will it contract neurodermitis?

The pedigree analysis is followed by a capsule introduction to medical statistics and general genetics. You learn that childbearing is always a risk: 3 to 5 % of all newborns have a congenital

[3] the same as above
[4] quotation of a genetic counseling session

defect. This is the “base risk” assigned to every pregnant woman. It resulted from the transfer of general statistical frequencies to individuals now conceptualized as subject to personal risk. The statistical observation, that on average one out of 200 newborns have a heart defect has become the individual risk of 0,5% for every pregnant woman to bear a child with a heart defect. Thinking in this way, one also runs a risk in using a bathroom. There is a 5% chance of a slip on the smooth tiled floor, resulting in a severe or even lethal injury.

Some counselors explain chromosomes and genes using the figure of a tape cassette: “Everything that is in or on us is in some way stored here.”[5] Every human being has 46 chromosomes, half from the father, half from the mother. After fertilization, cells divide, and a mistake might creep into this process. Then, cells with a surplus chromosome reproduce. Ah, you think, putting your hand on your belly that sometimes rumbles sensitively, I’ve already learned that at school. What does it have to do with me expecting a child? The counselor continues: if there are three #21 chromosomes, it is the genetic indication of Down’s syndrome. Didn’t your test deal exactly with that? Children with Down’s syndrome are said to be mentally retarded and developmentally hindered.

Thinking to yourself, you remember seeing a woman on the street sometimes, accompanied by a child who looked peculiar, but seemed to be of good spirits. In any case, it is difficult to raise a child, and you are in the middle of your studies. What would you do with a handicapped child? It cannot go to a normal school and needs intensive care. Then, you might not be able to finish your studies…

You listen as the counselor continues talking about chromosomes, amniotic fluid, low abortion risks, cells, gonosomal aberrations, trisomies, and that the presence of Down’s syndrome can be detected with that method. But she cannot decide if an abortion is the right choice for you. Normally, an amniocentesis is performed from the 15th week of pregnancy. For the result, one has to wait at least 2-4 weeks, which means, that you would be pregnant in the fifth month. The fact that an abortion after the sixteenth week of pregnancy is an induced stillbirth, starting with labor, often lasting many hours or days, and that a living fetus might result, is frequently not detailed. During the week following the counseling session, you have to decide whether you will undergo the amniocentesis.

You, Beate, have an independent mind and possess a healthy skepticism. If all the talk about the risks of anomalies and retardation has not worn you down or undermined your self-confidence, you might ask: What’s the good of all this? What has a test result to do with the child I am expecting, and whose growth I can more and more certainly feel? What can really be known about your daughter Anna, when her chromosomes have been counted? A chromosomal count can say nothing about the concrete character of Anna; the counselor can only make a prognosis. Statistical deviations from biological norms are translated into specific medical judgments like “mentally retarded” or “reduced life expectancy”. The relationship between the biotechnical lab result on chromosomes and a predicted medical condition in a certain individual is only a statistical correlation, nothing more. It doesn’t really say anything about your child. If your child turns out to be more easygoing or difficult, successful in school studies or more talented in practical arts, how your life together will turn out - genetic counseling has nothing to say. The counselor’s education only equips her to analyze probability profiles of prenatal populations.

If you don’t have any questions, the hour and a half introduction to genetic thinking comes to an end. You will receive a letter containing the most important points of information. It is designed to help you make a decision. In case you decide to undergo the examination - as more and more women being pregnant and on top of that aged thirty five or older do, because they think one should accept every medical offer to avoid any possible complication - and the chromosomes deviate from the norms, you will again be referred to the friendly but helpless expert, the genetic counselor. In contrast to other clinical sessions, the physician is also removed from any healing function; her task

[5] quotation of a genetic counseling session

is only to impart the future possibilities of test results. She will explain the lab results in detail, and you can then decide to bear your child for death, a child that is not yet there, and your reasons would be based on laboratory findings correlated with future possible and abstract conditions. Or you can choose to carry the pregnancy to term and expect a risk carrier: a child already tested, diagnosed, classified and with a prognosis even before it is born. In this situation, there is no possibility to do something on your own, something independent of the medical system, which is good and makes sense.

What I am especially interested in, Dear Beate, is the question: What has genetic counseling done to you? Even though nothing could be said about the concrete child you expect, after all that scientifically-based information, you cannot just tear up the letter from the counselor and wait for your child.

Your fears and anxieties during pregnancy, your queasiness when you think of caring for a handicapped child, belong to a reality radically different from the professional talk about genes and chorion villi. As a student, you are probably accustomed to encounter scientific concepts and you have already learned to find yourself placed in high risk groups and on statistical curves. You may want to inform yourself, while preserving a skeptical distance. But the counseling will leave an irremovable link between the general talk about risks, birth defects, rising statistical curves, and your invisible insides. The points of contact for this newly created relationship between your conscious person and your body are the physical tests joined to the scientifically-inspired informational bias, that is, the diagnostic procedures interpreted by technological care. The immediate result: misplaced concreteness given to statistical constructs.

Tests during pregnancy, now a routine procedure, have led you into a situation where you can only chose between given institutional options. Genetic counseling asks you to make a decision on the basis of a reference system heterogeneous to your experience. The more understanding and compassionate the counselor encourages you to acquire and embrace the scientific information in order to increase your autonomy, the more she misplaces you. You are torn out of an experienced, intensely haptic and meaningful context, and then left to orient yourself with the statistical terms and arbitrary constellations of an abstract sphere.

Dear Beate, I hope you are satisfied with my reply. Next time, we should think about the question what makes so many of your friends lose their self-confidence and believe in medical monitoring as soon as they expect a child. Why not ask them? Best wishes to George, and, of course, to little Anna.

Write soon,

Yours