

Legislation on Genetic Diagnosis: Comparison of South Korea and Germany - With Focus on the Application and Communication Structure -

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ABSTRACT : This article explores the questions regarding PND and PID, especially the concrete legal conditions for the justification of PND and PID. As such, the German law stipulating PND and PID in a very concrete and detailed manner is introduced and explained in comparison with the corresponding South Korean law. The South Korean Bioethics and Biosafety Act (BBA) stipulates various types of gene testing and does not demonstrate a delicate sense of each type of gene testing. In contrast to the South Korean regulation, in Germany, there exist specific regulations for genetic counseling. Especially in the case of PND, GEKO stipulates the process of genetic counseling very concretely, based on GenDG. In the case of PND and PID, it is important that the people concerned understand the meaning of testing in various angles, and restructuralize it by combining it with their own values as the diagnosis is directly combined with pregnancy/abortion, which influences the whole life of a woman (and her partner). In this context, the South Korean BBA needs to be amended as soon as possible. The sections on informed consent also need to be amended to make them more concrete. Furthermore, guidelines for concretizing the regulation of BBA need to be continuously formulated and developed.

Key words : Genetic Diagnosis, Gene Testing, PND, PID, Bioethics, Bioethics and Biosafety Act, Gendiagnostikgesetz

INTRODUCTION

The development of bioscience has enabled the genetic diagnosis of an unborn human life. The diagnosis as such is classified based on its target: PND (prenatal genetic diagnosis) and PID (preimplantation genetic diagnosis). PND is the diagnosis of the embryo and fetus ‘during pregnancy’ (i.e., ‘in the uterus’), and PID is the diagnosis of the embryo *in vitro*, produced via IVF (*in-vitro* fertilization) and not yet implanted in the uterus of a woman. The legal validity of the genetic diagnosis can be studied in two directions. On one hand, it has been argued the diagnosis itself could be allowed, especially in the context that the

selection of the embryo or fetus and abortion are permitted according to the diagnosis result, and if permitted, in which range it can be permitted. On the other hand, it has been discussed under which conditions genetic diagnosis could be justified in the sequence of genetic diagnosis, and how the concrete conditions should be legalized.

In South Korea, the Bioethics and Biosafety Act (BBA) stipulates “genetic testing,” and the sections of BBA on genetic testing states the specific genetic disease whose identification justifies the genetic diagnosis of an embryo and fetus in a so-called ‘positive’ way. In exploring the questions regarding PND and PID, this paper will be limited to the consideration of the concrete legal conditions for the

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justification of PND and PID. Such considerations could contribute in some way to the South Korean legal policy of genetic testing in BBA. Towards such end, the German law stipulating PND and PID in a very concrete and detailed manner will be introduced and explained in comparison with the corresponding South Korean law.

Macroscopic Structure of Legislation

1. Applicable law

1) Bioethics and biosafety act in South Korea

In South Korea, genetic diagnosis is regulated by BBA. Titled “Gene Therapy, Testing, Etc.,” Chapter 6 of BBA 2013 contains sections (sections 49–53) regarding the subject of gene testing (genetic testing institutions), the purpose and scope of gene testing, the informed consent of the testee, the management of records and information concerning gene testing, and the provision and discarding of the materials for gene testing. Especially, section 50(2) sets limits to the scope of gene testing conducted on an embryo or fetus for the purpose of diagnosing a genetic disease.

2) Human gene testing act and embryo protection act in Germany

In contrast with the expansive regulation of BBA, which contains a wide range of bioethical issues, including the production of and research on embryos and the research on human materials and gene therapy, there exists an act specifically on gene testing in Germany. The Human Gene Testing Act (Gesetz über genetische Untersuchungen bei Menschen (Gendiagnostikgesetz: GenDG)) was promulgated on July 31, 2009 and went into effect on February 1, 2010. The section on the definition of gene testing in BBA states that the only gene testing specimens allowed by BBA are “human materials” (§2 Nr.15 GenDG); likewise, GenDG

stipulates gene diagnosis with living people, embryos, and fetuses as objects, and related matters. This is clear from the full name of GenDG and from §2① GenDG, which stipulates the range of application of GenDG, but while the same regulation of BBA is applied to both PND and PID in South Korea, the German GenDG is not applied to PND because gene testing on embryos and fetuses in GenDG is limited to that on the embryo or fetus “during pregnancy” (§2① GenDG). The object of PID (i.e., embryo *in vitro*) is actually the object of regulation of the Embryo Protection Act (Gesetz zum Schutz von Embryonen (Embryonenschutzgesetz: ESchG)), which defines the embryo as the fertilized, viable human egg formed at the time of nuclear fusion (§8① ESchG), but this was subsequently disputed through the amendment of ESchG in 2011, prohibiting PID, especially of §1① Nr.2ESchG, prohibiting artificial fertilization for any purpose other than pregnancy, and of §2① ESchG, prohibiting the usage of an extracorporal human embryo for any purpose other than promoting its preservation. Before the 2011 amendment, ESchG did not have any section clearly aimed at regulating PID. On July 6, 2010, the German Supreme Court (BGH) ruled that §1① Nr.2ESchG and §2① ESchG cannot be applied to PID via blastocyst biopsy, and to the subsequent investigation of the extracted pluripotent trophoblast aimed at the discovery of a serious genetic damage (BGH, Urteil vom, 6 July 2010, 5StR 386/09). After this judgment, a new section on PID was enacted in ESchG (§3a ESchG) by the PID Regulation Act (Gesetz zur Regelung der Präimplantationsdiagnostik) on November 21, 2011. Additionally, GenDG set up an interdisciplinary, independent committee for genetic diagnosis (Gendiagnostik-Kommission: GEKO) at Robert Koch Institut, a federal institute under the German Federal Ministry of Health (§23① GenDG). The committee’s primary assignment is to establish concrete guidelines concerning the various issues regulated by GenDG. It would be of great signifi-

cance for the debate on the analysis and policy of BBA to introduce the contents of the GEKO guidelines concretely in this paper.

2. Applicable scope

1) One act with an extensive scope in South Korea

BBA defines gene testing as “a test conducted to obtain genetic information from a human material for identifying an individual or for preventing, diagnosing, or treating a disease” (§2 Nr.15 BBA). This means that BBA stipulates two types of gene testing: on one hand, gene testing for diagnosis and therapy (i.e., for medical purposes), and on the other hand, gene testing for the identification of an individual. Especially, according to the authoritative interpretation of the South Korean Ministry of Health and Welfare (KMH) and Korea Centers for Disease Control and Prevention (CDCP), gene testing for research purposes is included in the types of gene testing allowed by BBA. <Guidelines on the Administration of Institutions Relating to BBA>, drawn up by KMH and CDCP, clarifies that gene testing for research purposes is allowed if it is approved by the Institutional Review Board for Bioethics (IRB) (KMH & CDCP, 2013).

2) Different legislations in Germany

Unlike the South Korean GenDG, the German GenDG stipulates gene testing with two major criteria, and has sections with specific and concrete contents. On one hand, the German GenDG classifies the subjective purposes of gene testing into “medical purposes” (Chapter 2 of the German GenDG) and “descent determination” (Chapter 3 of the German GenDG), and stipulates the conditions for the justification of each type of gene testing. On the other hand, the German GenDG cites two areas (i.e., the insurance and work/labor areas) where gene testing and the use of its results or of the information obtained from it are basically

not permitted. Especially, it is worth noting that gene testing and the analysis and usage of gene samples and of the information obtained from gene testing conducted “for research purposes” are not included in the range of the German GenDG’s regulation (§2②Nr.1 GenDG), in contrast to the authoritative interpretation of BBA in South Korea. Furthermore, while gene testing conducted to determine descent is regulated by BBA in South Korea, especially by §2 Nr.15 BBA, which determines the term of gene testing, the German GenDG stipulates that its provisions do not apply to genetic research and analysis conducted as part of the criminal process, and to the handling of genetic samples and data within the same process (§2②Nr.2 GenDG). In Germany, gene testing for evidence and identity verification is stipulated by the Criminal Process Act (Strafprozessordnung: StPO), especially by §81g, §81h StPO. The regulation of gene testing in the German StPO has many implications on the South Korean situation, where the attempts to legalize matters concerning the use of genetic information and the conduct of gene testing in the criminal process have consistently failed (for a fuller discussion of these issues, Cho, 2007).

Microscopic Structure of Regulation

1. Purpose of diagnosis

1) Enumeration system in South Korea

As mentioned earlier, in South Korea, the genetic diagnosis of an embryo or fetus is allowed only for the identification of specific genetic diseases. This is called a ‘positive way of stipulation’. BBA stipulates that gene testing may be conducted on an embryo or fetus “only for diagnosing muscular dystrophy or any other hereditary disease specified by presidential decree” (§50② BBA). Based on this section of BBA, the Enforcement Decree of BBA (ED BBA) enumerates 62 genetic diseases (§21, Schedule3 ED BBA),

and based on Nr. 63 of Schedule 3 ED BBA, the Notice from the Secretary of the Korean Ministry of Health and Welfare (KMH) enumerates 91 genetic diseases. That is, there are 153 genetic diseases in all that are allowed to be diagnosed through gene testing on an embryo or fetus.

2) Stipulation by subjective purpose and type of action in Germany

In contrast to the South Korean regulation, in Germany, different laws apply to PND and PID, as mentioned earlier. PND is stipulated by the German GenDG, particularly by §15 in the section on gene testing for medical purposes (Chapter 2 of the German GenDG), titled “Prenatal Gene Testing.” PND is not always conducted for medical purposes, but §15 states that prenatal genetic diagnosis can be conducted only for medical purposes. Thus, the range of the permitted PND is limited to diagnosis for medical purposes. §15① GenDG states that PND can be conducted if ① the diagnosis is aimed at determining the specific genetic characteristics of the embryo or fetus that affect its health during the mother’s pregnancy or after birth, according to the generally acknowledged state of science and technology, or ② the treatment of the embryo or fetus is provided with a drug, whose effect is influenced by certain genetic characteristics. These purposes are the only subjective purposes that are stipulated as the criteria for the decision to permit the diagnosis. If the intention of the diagnosis is outside the range of these purposes, imprisonment up to one year, or a fine, shall be imposed on the one who conducted it (§25① GenDG). Furthermore, the German GenDG defines gene testing as a superordinate concept for “gene analysis” and “prenatal risk evaluation,” and states that the terms *gene analysis* and *prenatal risk evaluation* include the assessment of the results (§3①a,b GenDG).

Regarding PID, §3a① ESchG stipulates that anyone who analyzes an embryo *in vitro* before its intrauterine transfer (i.e., anyone who conducts PID) will be punished

with imprisonment up to one year, or a fine. This means that ESchG stipulates that the fundamental principle is to prohibit PID penally. ESchG, however, also stipulates the conditions for the exceptional permission of PID, as BGH clarified in its judgment that PID can be permitted when it is conducted for medical purposes, such as for the discovery of a serious genetic damage. ESchG prescribes the medical purpose of diagnosing the health condition of the embryo, including the possibility that it has a genetic disease, as the necessary subjective condition for the permission of PID. §3a② ESchG states that PID is not illegal in the following instances: ① if there is a high risk of having a serious genetic disease on the part of the descendants due to the genetic disposition of the woman from whom the ovum originates, or of the man from whom the sperm originates, or both, and if the diagnosis is conducted according to the generally recognized state of medical science and technology, and with intent to induce pregnancy; or ② if the diagnosis is conducted to determine a serious damage that will very likely lead to the death of the embryo, or to a miscarriage.

2. Genetic communication

1) Informed consent

One of the most important conditions for justifying the conduct of PND and PID is informing the pregnant woman or the woman from whom the ovum originates that she will be subjected to PND or PID, and obtaining her consent to be subjected to gene testing.

(1) Stipulation of core principles in South Korea

§51 BBA stipulates that the informed consent of the subject must be obtained before the conduct of gene testing. Above all, the information should be obtained by a gene testing institution (§51⑥ and §51① BBA). Especially in the case of PND and PID, only medical institutions are

permitted to conduct gene testing, according to §50③ BBA; therefore, the person who will obtain the information concerning PND and PID should be a medical personnel. The medical personnel should “sufficiently” explain “the objectives of the gene testing to be conducted, the method to be used for it, and its expected results and significance” to the testee (§51⑥ BBA), and should obtain the written consent of the testee with regard to the following matters: ① the objectives of the gene testing to be conducted; ② the management of the material for testing; ③ the withdrawal of consent; ④ the protection of the subject’s rights and of the information obtained from the test; and ⑤ other matters specified by the Ordinance of the Ministry of Health and Welfare (§51① BBA). A person who conducts gene testing (actually, who extracts a gene sample from a person to be used for gene testing without written consent from such person) shall be punished by imprisonment with prison labor for not more than one year, or by a fine not exceeding 20 million won (§68 Nr.11).

(2) Specification of informed consent in Germany

- **Explanation (giving information):** §9① GenDG states that the responsible medical person should provide an explanation before conducting PND. The object of the explanation by §9① GenDG is the person concerned, and §15① GenDG concretizes it in the context of PND; thus, the information needs to be given to the pregnant woman in the case of PND. The relevant information must be given to the pregnant woman before obtaining her consent, and the person concerned must be given “a reasonable period of consideration” (eine angemessene Bedenkzeit) of whether to give her consent (§9① S.2 GenDG). This stipulation sets the foundation for the concept of medical autonomy (i.e., the essential basis of the informed consent) in the area of gene testing. Furthermore, the realization of

the autonomy of the testee premises the explanation of the relevant matters that have a decisive effect on whether to consent to the diagnosis. As such, the German GenDG states that the “nature, significance, and implications of gene testing” must be explained (§9① GenDG), and the contents of the explanation need to be documented before the conduct of gene testing (§9③ GenDG). Additionally, in contrast to the Korean Civil Law, the German Civil Law (Bürgerliches Gesetzbuch: BGB) stipulates a “medical contract” as a special form of contract, and has sections especially on obtaining the informed consent of the testee (§630d, §630e BGB). The sections on obtaining the informed consent of the testee state that the relevant information must be given “in a timely manner” (rechtzeitig) so that the patient can make a decision on whether to consent to the procedure “with prudence” (wohlüberlegt). As mentioned earlier, the South Korean BBA states the duty of providing a “sufficient” explanation in the case of gene testing (§51⑥ BBA), but does not fully specify the need to understand the explanation as a way of realizing one’s autonomy through the reflective decision of the testee. Therefore, examples of German legislation with regard to obtaining the informed consent of the testee in the area of medicine imply much in terms of determining the improvement direction of the South Korean law policy.

- **Consent:** §15① GenDG states that §8① GenDG applies to the consent to PND. The consent to be subjected to gene testing and to the procedure for obtaining the necessary gene sample must be given to the medical person responsible for the conduct of the testing “expressly” (ausdrücklich) and “in writing” (§8① S.1 GenDG). Further, the consent includes not only the decision to subject oneself to PND but also the decision on whether to require the medical institution that will conduct the procedure to provide oneself with the results of the procedure, and if so, on the method of provision to be employed, or to allow the

obtained data to be discarded. Also, gene analysis can be done only when there is evidence that the testee gave his/her consent to such (§8① S.2 GenDG).

2) Genetic counseling

(1) No legislation in South Korea

In South Korea, genetic counseling is not structuralized in medical practice (Kim, 2011a). First of all, no legislation for genetic counseling exists, and no authorized education system for genetic counseling specialists or certification program (Kim, 2011a). There are genetic-medical clinics, but genetic counseling conducted by genetics specialists is quite rare. It is sometimes delegated to the nurses in the related departments, or even to the employees of laboratories (Kim, 2011a).

(2) Concrete legislation in Germany

In Germany, there exist specific regulations for genetic counseling. Especially in the case of PND, GEKO stipulates the process of genetic counseling very concretely, based on GenDG. Concerning PID, ESchG stipulates that counseling about the medical, psychological, and social results of gene testing with embryonal cells must be done before testing (§3a③ S.1 Nr.1 ESchG). Below, the German regulation of genetic counseling as such will be introduced, especially focusing on the sections of GenDG that stipulate genetic counseling in detail.

- **Claim to counseling:** GenDG expressly states that a pregnant woman can avail of “genetic counseling” service with regard to PND. Genetic counseling as such has many implications on the South Korean BBA, which does not mention genetic counseling at all. A pregnant woman can take part in genetic counseling as stipulated by §10 ② and §10 ③ GenDG before PND and after obtaining the test results. Furthermore, a pregnant woman can be additionally

provided with counseling, as stipulated by §2 of the Act on the Prevention and Management of Pregnancy Conflict (Gesetz zur Vermeidung und Bewältigung von Schwangerschaftskonfliktgesetz (Schwangerschaftskonfliktgesetz: SchKG)) (§15③ GenDG). SchKG states that every woman/man has the right to be informed anonymously or to obtain counseling on family planning issues and on all questions directly or indirectly related with pregnancy from a counseling body dedicated to such matters (§2① SchKG).

- **Process of counseling:** GEKO more concretely stipulates the process of genetic counseling with regard to PND. The GEKO guidelines can be summarized as follows: the genetic counseling regulated by GenDG is a process of ‘understanding-consideration-choice-decision’ on the part of the client, and a process of guaranteeing the client’s ‘autonomy’ for his/her independent decision, and the right ‘not to know’ in the same context, on the part of the counselor. According to the GEKO guidelines (GEKO, 2011a), genetic counseling in the process of PND enables the person concerned to understand the medical-genetic meaning of the test or its results, to ponder the options and to choose from among them, and to decide for him/herself. Particularly in the case of genetic counseling provided ‘before’ the conduct of gene testing, GenDG states that “a reasonable period of consideration” — as in the case of informed consent — must be allowed until testing (§10② GenDG). This guarantees the counseling needed to set the basic condition for intact decision-making, as in the case of the explanation of the procedure.

- **Structuralization and specialization of reflection:** The realization of the client’s medical autonomy through the process of counseling is fundamentally possible only when the process of the client’s reflection on her/himself and on the future life of the baby is structuralized (i.e., multidirectional consideration is possible when the client

ponders the indication of gene testing and the discussions about medical, psychological-social, and ethical matters (GEKO, 2011a), and hereby understands the meaning of gene testing from an objective viewpoint). For this, it is important, above all, that the client views her/his own situation accurately before discussing how to accept it subjectively. This means that the client needs to be informed of the average risk of developing a health disorder on the part of the newborn infant so that the client would not make a decision solitarily when facing the prediction of the fetus's genetic disorder, etc (GEKO, 2011a). Further-more, the genetic counseling must be internally structuralized in such a way that the specialists of various areas respect the client's "values and religion" and "psychological-social situation" (GEKO, 2011a), and simultaneously help the clients through the dialectic synthesis of their own values and the psychological and ethical views related with genetic problems. In this context, GenDG expressly states that another specialist can be involved in matters concerning obtaining the consent of the client (§10③ S.3 GenDG).

CONCLUSION

As pointed out earlier, the South Korean BBA stipulates various types of gene testing and does not demonstrate a delicate sense of each type of gene testing. Especially in the case of PND and PID, it is important that the people concerned understand the meaning of testing in various angles, and restructuralize it by combining it with their own values as the diagnosis is directly combined with pregnancy/abortion, which influences the whole life of a woman (and her partner). In this context, the South Korean BBA, which does not have any stipulation on genetic counseling, needs to be amended as soon as possible. Especially, the qualification system for professional counseling and educational programs are the essential premises for guaranteeing the conduct of genetic counseling not only scien-

tifically but also "compassionately and sensitively" (Jun & Anderson, 2011). The sections on informed consent also need to be amended to make them more concrete. Furthermore, guidelines for concretizing the regulation of BBA need to be continuously formulated and developed. Biotechnology is always in progress, and the South Korean BBA cannot reflect all the relevant practical situations without the help of the guidelines as such. The system of making and developing guidelines needs to be structuralized concretely in the South Korean BBA. The South Korean BBA thus still has a long way to go, especially with regard to genetic diagnosis.

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