

# Directive or non-directive genetic counselling – Cutting through the surface

Pekka Louhiala  
Hjelt Institute  
University of Helsinki, Finland  
[pekka.louhiala@helsinki.fi](mailto:pekka.louhiala@helsinki.fi)

Veikko Launis  
Department of Clinical Medicine  
University of Turku, Finland  
[veikko.launis@utu.fi](mailto:veikko.launis@utu.fi)

## Abstract

*A case is first presented in which a woman carries a fetus with Turner syndrome. Thereafter, the development of non-directiveness to a standard in genetic counselling is described, and the concepts of directive and non-directive counselling are analysed critically. Next, the treatment of Turner syndrome in medical texts and websites is examined in detail. Finally, it is argued that the concepts of directive and non-directive counselling are anything but clear and, in particular, not helpful in answering the question concerning decision making in the case described and in other relevantly similar cases.*

**Key Words:** Turner Syndrome; Genetic Counselling; Directive; Non-Directive

## Introduction

A 36 year old woman is 17 weeks pregnant and has had an ultrasound examination and amniocentesis to detect possible chromosome aberrations in the fetus. The ultrasound was normal, but the amniocentesis revealed that the fetus has only one X-chromosome and has Turner syndrome. How should the woman be counselled?

The standard answer to this question is that the woman should receive *non-directive counselling*, which is generally understood as giving morally neutral information about the condition and the alternatives the client has. The term is, however, problematic and often left unexamined. According to Angus Clarke, “continual reference to an unexamined ideal of ‘non-directiveness’ may have mislead professionals into providing clients with information but then backing away from supporting them in their decision-making, in case this is interpreted as an attempt to influence their decision” (Clarke, 2007).

In this article, we first describe the development of non-directiveness to a standard in genetic counselling. Second, we examine the concepts of directive and non-directive counselling. Third, we study in detail several medical texts and websites that deal with Turner syndrome. We then argue that the concepts of directive and non-directive counselling are anything but clear and, in particular, not helpful in answering the question concerning decision making in the described case and in other relevantly similar cases.

## Material and Methods

The study material is the existing scientific literature about genetic counselling in general and Turner syndrome in particular. The research method is comparative discourse analysis and systematic literary review. The theoretical basis of comparative

discourse analysis lies in social constructionism that refers to the way that the reality is perceived as constructed through the language. Comparative discourse analysis is critical and it is purported to make power relations and hidden value-judgments visible and question them by introducing alternative discourses.

An analysis of medical texts and web sites concerning TS is performed, identifying potential patterns of shared and divergent values and positions taken by them. Then, the opinions are analysed for patterns of shared and divergent values.

## Non-Directiveness as a Standard

Genetic counselling is “what happens when an individual, a couple or a family asks questions of a health professional [the genetic counsellor] about a medical condition or disease that is, or may be, genetic in origin” (Clarke 1994). It may cover many different activities, but at its core it is a process of communication (Elwyn et al., 2000). In the words of Angus Clarke, a medical doctor and clinical geneticist: “We dispense words, not tablets” (Clarke, 1997a).

In the 1960’s and 1970’s genetic counselling was often quite *directive* in the sense that counsellors did not hesitate to express their own moral views about what their clients should do. The outcome and success of genetic counselling was measured, for example, in terms of whether the clients had accepted the counsellor’s conclusions or whether the outcome of the reproductive choice was the one suggested by the counsellor (Mitchie et al., 1997). C.O. Carter, one of the pioneers of clinical genetics, encouraged parents with low or moderate risks (less than 1 in 20) by saying that “in your place I would be prepared to take the risk” (Carter et al., 1971). Another paper, also from the early 1970’s, opens by stating: “Genetic counselling is preventive medicine and should be so regarded” (Leonard, Chase and Childs, 1972).

These comments not only reflect genetic

counselling, but also the prevalence of paternalistic thinking in the medicine of those days. The concept of non-directiveness was, however, codified already in 1974 by a committee of the American Society of Human Genetics to include the provision of full information and help to individuals or families to make their own autonomous decisions (Fraser, 1974). Gradually, non-directiveness became a recognised standard in several official reports on both sides of the Atlantic.

Why did non-directiveness later become such a standard or ethical cornerstone of genetic counselling? Apparently for three reasons. Firstly, genetic counselling developed as an activity partly as a reaction to the abuses of human genetics in Europe and North America during the first half of the 20th century (Clarke 1997b). Genetic counsellors generally wanted to avoid being accused of “playing god” or practising eugenics or genetic discrimination of any sort.

Secondly, historically, the development of genetic counselling coincides with the general improvement in patient autonomy. The doctor-patient relationship had long been viewed as a relationship of more or less blind *trust* instead of as an equal partnership with symmetrical knowledge, power, and autonomy because a paternalistic view of medicine was assumed (O’Neill, 2002).

Thirdly, an emotional explanation has been suggested that non-directiveness may help the professionals to keep an emotional - and perhaps also moral - distance from the decisions made by clients and also help them to maintain distance from the politics of second trimester abortion. According to Bosk (1992):

“Genetic counselling as a service is generally a matter of transferring information to individuals who request it, and then leaving those individuals alone to make the tragic choices based on that information.”

#### **The Nature of Directive and Non-Directive Counselling**

Although the terms *directive* and *non-directive counselling* have been commonly used for decades, it is surprising that they have seldom been defined. The following definition for directiveness has been provided by Seymour Kessler, an experienced researcher and educator in genetic counselling: “Directiveness in genetic counselling is a form of persuasive communication in which there is a deliberate attempt - through deception, threat or coercion - to undermine the individual’s autonomy and compromise his or her ability to make an autonomous decision.” (Kessler, 1997)

Although the concept of directiveness remains vague in other writings, it usually seems to imply that the counsellor more or less deliberately gives advice or directions in a non-neutral fashion. Such directiveness does not necessarily mean deception or threatening, and resorting to coercion

may even be considered to exceed the limits of what we normally mean by persuasive *communication* (Mill, 1978).

Non-directiveness is a vague concept as well. Kessler has argued that directiveness and non-directiveness have too often been understood as opposite sides of the same coin (counselling is taken to be either directive or non-directive), while a more fruitful understanding of these terms might be to see them as “extremes of a more or less normal distribution of transactual possibilities in counselling sessions” (Kessler, 1997). Elwyn et al. (2000) have remarked that there is an inherent difficulty in separating the *giving of information* from the *giving of advice* [our italics]. The former is acceptable in the context of non-directive counselling while the latter is not.

We would like to add one more point: it is possible that the relationship between directiveness and non-directiveness is not symmetrical. While it is easy to imagine what fully directive counselling would be like in practice, it may be that fully non-directive counselling is not at all possible (see our discussion below).

In an empirical study on directiveness in genetic counselling, the following operative definition for directiveness was provided:

“Directions or advice that the counsellor suggests to the client in regard to specific behaviours or making decisions. Directions or advice about the client’s views, attitudes or emotions” (Mitchie et al., 1997).

Of course, this definition does not make full justice to the complex phenomenon, but in empirical research definitions like this are necessary.

The researchers recorded counselling sessions and classified the statements of the counsellors into the categories advice, evaluation, and reinforcement. An example of a piece of advice would be “It’d be sensible if you spoke to Michael and Carol about this”, and an evaluation would be “That is what we would consider quite a high risk”, while reinforcement would be “I think you’ve made the right decision”.

According to the analysis, all consultations contained at least two directive statements. Perhaps surprisingly, the counsellors who had received counselling training were more directive than those who had not received it. The counsellors were also asked to rate themselves on a directiveness scale from 0 to 6, and none of them rated themselves at the extremes.

However, the degree of directiveness varies according to the seriousness of the condition in question. According to a study by Marteau et al. (1994), a directive approach was adopted by many counsellors when the condition was lethal (for example, anencephaly) or relatively minor (for example, cleft lip). Nondirective approaches were related to late-onset disorders (such as Huntington’s disease) and disorders with variable expression (such as sickle cell disease).

Other studies have addressed the differences between counsellors as regards the degree of directiveness. Males have been shown to be more directive than females (Wertz and Fletcher, 1988) obstetricians more directive than geneticists, and the latter more directive than

genetic nurses (Marteau, Drake and Bobrow, 1994). It is, however, possible that these reported differences in attitude reflect an awareness of the “professionally correct” responses on questionnaires rather than real differences in practice (Clarke, 1997b).

Non-directiveness is not necessarily the approach the clients want or feel comfortable with. In a Finnish study, for example, families having received genetic counselling were asked whether they wanted to hear only the facts or also the physician’s advice about having more children, and 42% of the respondents expressed a wish to hear both (Somer, Mustonen and Norio, 1998). In a British study it was found that the more neutral the counsellor was perceived to be, the higher the counsellee perceived his or her own risk to be (Mitchie et al., 1997). When the counsellor was perceived to be non-directive, he or she was perhaps also perceived to be concealing bad news. In addition, it has been found that the measures of directiveness are not associated with counsellee satisfaction with information, mood, or the extent to which counsellee expectations are met.

Although non-directiveness is a generally accepted goal, some counsellors have expressed their awareness of a discrepancy between non-directiveness and the needs of the people they work with (White 1998). Their experience of genetic counselling is that a completely neutral stance is often regarded as cold and unhelpful. In addition, the expectation of unconditional support of the clients’ decision—in particular if it is felt to be unethical—can be highly stressful for the counsellor.

Occasionally, a non-directive approach may even be ethically inappropriate. It may be in the interests of other family members that the client discloses information about her/himself if the family members are at risk of developing a genetic disorder or having a child affected by a genetic disorder. In such cases it may be the duty of the counsellor to try to persuade the client to disclose such information (Elwyn et al., 2000).

#### Turner Syndrome in Medical Texts

To get a picture of the ways Turner syndrome (TS) is described in medical texts, we examined seven paediatric textbooks (Rudolph and Hoffman, 1987; Avery and First, 1994; Campbell and McIntosh, 1998; McMillan et al., 1999; Behrman, Kliegman and Jenson, 2000; Saenger, 2008, Kliegman and Nelson, 2011), five review articles (Saenger et al. 2001; Ranke and Saenger, 2001, Sybert and McCauley, 2004; Bondy, 2007, Pinsker, 2012). In addition, we examined three websites, one created by and for medical professionals, one created by medical professionals for the general public, and one created by a TS organisation ([www.emedicine.com/ped/topic2330.htm](http://www.emedicine.com/ped/topic2330.htm); [www.nhs.uk/conditions/turners-syndrome/Pages/Introduction.aspx](http://www.nhs.uk/conditions/turners-syndrome/Pages/Introduction.aspx); [www.turnersyndrome.org/](http://www.turnersyndrome.org/)).

It should be kept in mind, however, that the texts were written for different audiences and purposes: medical texts for medical professionals

taking care of people with TS, and general texts for the general audience.

Typically, the textbooks provide long lists and tables about the clinical features found in individuals with TS. Almost always the clinical picture is described with negative terms. Rudolph and Hoffman (1987), for example, open the description with a list of such features:

“[TS] ... is characterized by a female phenotype, short stature, sexual infantilism, streak gonads, and a diversity of associated somatic anomalies” (Rudolph and Hoffman, 1987: 1536).

They then provide a list of those anomalies and estimates of their prevalence and a discussion on the development of the characteristic physical features during childhood and puberty. Hormonal therapy is commented shortly but no reference is made to quality of life issues. The most recent textbook (Kliegman and Nelson, 2011) is not different in this respect.

Behrman, Kliegman and Jenson (2000) begin by describing the discovery of the syndrome and its pathogenesis. The clinical manifestations of TS are then pictured lengthily and aspects of growth hormone and oestrogen therapy are discussed in detail. The need of psychosocial support is mentioned as an integral component of treatment and the value of patient organisations is recognised. In the end of the chapter the life of TS adults is briefly described, again in terms of medical problems.

Only one of the seven textbooks briefly mentions that girls with TS have a normal life span (Campbell and McIntosh 1998). In one textbook the word “individual” is used to refer to people with TS (McMillan et al. 1999); the rest use the word “patient”. The most comprehensive textbook review (Saenger, 2008) ends by stating that with “the intervention strategies described, girls and women with Turner syndrome now - more than ever - have the capability of achieving their full potential.”

The review articles also provide long lists of problems that need medical attention. A major difference between the textbooks and the review articles is that the latter also mention the generally good prognosis: “most patients with Turner’s syndrome are socially well-integrated at all levels” (Ranke and Saenger, 2001), and “most adults with Turner’s syndrome report satisfaction with their lifestyle” (Sybert and McCauley, 2004). Two of the five articles do not comment on the problems of genetic counselling, but, in one article, the authors write: “It should be emphasized that individuals with TS can be healthy, happy, and productive members of society” (Saenger et al., 2001). None of the textbooks and only one review article mentions the uncertainty of the prenatal diagnosis. According to a Danish study, up to 30% of cases of TS diagnosed prenatally showed a normal karyotype at delivery (Pinsker, 2012, Gravholt et al., 1996).

Like the textbook and review articles, the websites created by medical professionals provide exhaustive lists of possible problems. The *NHS* website mentions that “girls and women with Turner syndrome will need regular health checks throughout their lives”, but, “however, most are able to lead relatively normal, healthy lives.” The *emedicine*

website notes that “[m]ost people live long and healthy lives, yet some are susceptible to a number of chronic conditions” and that the “overall prognosis for patients with Turner syndrome is good.”

Less surprisingly, the character of the *TS society* website is very different. In place of pictures of malformations, it contains smiling faces of individuals with TS. Instead of chromosome loss or aberration, it uses the term “chromosomal condition”. It is emphasised that “the majority of those with TS are healthy and well adjusted.” However, “all benefit from proper medical care, emotional support and careful screening for related conditions.”

#### Directive or Non-Directive Counselling?

According to the report of the Ethics Committee of the Royal Dutch Society of Physicians,

“directive counselling entails a revival of the classical role of the eugenically motivated ‘gatekeeper’—which might increase the resistance to genetic counselling” (de Wert et al., 2003).

The same report acknowledges, however, that, in some exceptional cases, directiveness can be allowed. The report refers to extreme risk situations in which there is a high risk of devastating harm to the future child. Criteria for acceptable directiveness in such situations are (1) that the counsellor stress that he or she is giving his or her personal opinion, (2) the counsellor tries to influence the client only by rational (non-coercive) persuasion, and (3) the counsellor discusses these exceptional cases with his or her team or asks for a second opinion.

Obviously, the term ‘directive’ is not used in the report in the sense Kessler defined it earlier (“through deception, threat or coercion”), but in a less extreme sense, referring to a situation in which the counsellor is, exceptionally, making value judgements about the situation.

It should be noted that our case example, in which a pregnant woman carrying a foetus with TS should be counselled, is very different from the situations considered in the Dutch report. The wording of the report seems to refer to extreme risk situations in which advice to *terminate* the pregnancy would be acceptable.

What, then, should be thought of non-directive counselling in our case? Is it morally desirable or even possible? Textbook and review articles do not serve as a good basis for non-directive information, because the focus of information in them is very often on negative descriptions of the syndrome. It is also possible that an attempt to be as non-directive as possible would mean adopting the kind of attitude that the clients would not wish in most cases. The least the counsellor should do to fulfil the counsellee’s moral and cognitive needs is to inform him or her about the ethical nature of the decision

(such as what kind of ethical values and reasoning is involved), as well as the possible consequences of it (Yarborough, Scott and Dixon 1989).

As to the second question, it has been suggested that non-directive counselling is, in principle, not possible. This is a direct result of the structure of the encounter between the counsellor and the client (Clarke 1991; Glover 2006). An offer of prenatal diagnosis, publicly organised and readily recommended both by the medical profession and national health care authorities, implies a recommendation to accept that offer, which entails a tacit recommendation to terminate a pregnancy in case of abnormality. The societally well-accepted offer of prenatal diagnosis is thus in itself a value statement, or can be read as such.

#### Concluding Remarks

A case presented in the *Hastings Center Report* in 1996 was similar to the case examined in this article (Case study, 1996). In a commentary, a genetic counsellor wrote:

“[I]t is my primary task to provide women and couples with the information they seek, so they can make informed decisions based on their values” (Punales, 1996).

This may sound simple, but it is far from it. As we have seen above, the information can be presented in various ways, without being incorrect as such. The Nuffield Council on Bioethics report (1993) on genetic screening points out that, “[i]n practice, a dialogue that helps an individual to explore the facts and issues in the context of his or her particular social and moral background is unlikely to remain completely neutral”. In addition, the counsellor’s attempt to be as neutral as possible may not be what the parents want.

Because the terms “directive” and “non-directive” are so problematic, it has even been suggested that they should be abandoned in the context of genetic counselling altogether (Wolff and Jung 1995). Even if the terms are accepted, most of what happens in genetic counselling takes place in the vast grey zone that can adequately be described neither as directive nor as non-directive (Oduncu 2002).

What can be said about the question posed in the beginning of this paper? How should the woman be counselled? She should, of course, receive adequate information about the medical facts of TS. However, these so-called facts have a social history and they are not as hard as they appear. In the words of Molewijk et al. (2003), “the facts have travelled a long, hidden and sometimes arbitrary journey before they are presented as ‘the facts’”. As our analysis shows, the ‘medical facts’ in the case of TS describe almost entirely the dark side of a life with the syndrome. But most of what life is about cannot be described in medical terms. Consequently, we think she should also receive adequate information about the lives of girls and women with TS. Sometimes, but only sometimes it might be appropriate to discuss the ethical nature of the decision to continue the pregnancy or abort the foetus.

#### Funding:

No external funding.

## References

- Avery, M.E., First, L. R. (1994). *Pediatric medicine*. 2nd ed. Baltimore (MD): Williams & Wilkins.
- Behrman, R.E., Kliegman, R. E., Jenson, H. B. (2000). *Nelson textbook of pediatrics*. Philadelphia (PA): WB Saunders.
- Bondy C. A. (2007). Care of Girls and Women with Turner Syndrome: A Guideline of the Turner Syndrome Study Group. *Journal of Clinical Endocrinological Metabolism* 92(1): 10–25-
- Bosk, C.L. (1992). *All God's mistakes: genetic counseling in a pediatric hospital*. Chicago/London: The University of Chicago Press.
- Campbell, A.G.M., McIntosh, N., (1998). *Forfar and Arneil's textbook of paediatrics*. New York (NY): Churchill Livingstone.
- Carter, C.O., Evans, K.A., Fraser Roberts, J. A., Buck, A. R., (1971). Genetic clinic follow-up. *Lancet* 1: 281–285. Case study: society's diseases. (1996). *Hastings Cent Rep* 26(3): 21.
- Clarke, A. (1991). Is non-directive genetic counselling possible?. *Lancet* 338: 998-1001.
- Clarke, A. (1994). *Genetic Counselling. Practice and Principles*. London/New York: Routledge.
- Clarke, A. J. (1997a). Outcomes and processes in genetic counselling. In P.S. Harper and A.J. Clarke (eds.) *Genetics, society and clinical practice* (pp. 179-200). Oxford: BIOS Scientific Publishers.
- Clarke, A.J. (1997b). The process of genetic counselling: beyond non-directiveness. In P.S. Harper and A.J. Clarke (eds.) *Genetics, society and clinical practice* (pp. 179-200). Oxford: BIOS Scientific Publishers.
- Clarke, A. (2007). Genetic counselling. In R.E. Ashcroft, A. Dawson, H. Draper and J.R. McMillan (eds.) *Principles of Health Care Ethics, Second Edition* (pp. 427-434). Chichester: Wiley.
- Daniel MS. Turner Syndrome. Medscape Reference. Retrieved on Dec 21 2013 from: <http://www.emedicine.com/ped/topic2330.htm>
- de Wert, G, ter Meulen, R., Mordacci, R., Tallacchini, M. (2003). *Ethics and genetics: a workbook for practitioners and students*. New York/Oxford: Berghahn Books.
- Elwyn, G, Gray, J., Clarke, A., (2000). Shared decision making and non-directiveness in genetic counselling. *Journal of Medical Genetics* 37:135-138.
- Fraser F.C. (1974). Genetic counselling. *American Journal of Human Genetics* 26:636–659.
- Glover, J. (2006). *Choosing children: genes, disability, and design*. Oxford: Clarendon Press, pp. 32—33.
- Gravholt, C.H., Juul, S., Naeraa, R.W., Hansen, J. (1996). Prenatal and postnatal prevalence of Turner's syndrome: a registry study. *British Medical Journal* 312:16–21.
- Kessler, S. (1997). Genetic counseling is directive? Look again [letter]. *American Journal of Human Genetics* 61:466–467.
- Kliegman R, Nelson W. E. (2011). *Nelson Textbook of Pediatrics*. Philadelphia: Elsevier/Saunders.
- Leonard, C.O., Chase, G. A., Childs, B. (1972). Genetic counseling: a consumer's view. *N Engl J Med* 287:433–39.
- Marteau, T., Drake, H. Bobrow, M. (1994). Counselling following diagnosis of fetal abnormality; the differing approaches of obstetricians, clinical geneticists and genetic nurses. *J Med Genet* 31:864–867.
- Marteau, T, Drake, H. Reid, Feijoo, M., Soares, M., Nippert, I., Nippert, P., Bobrow, M. (1994). Counselling following diagnosis of fetal abnormality: a comparison between German, Portuguese and UK geneticists. *Eur J Hum Genet* 2:96–102.
- McMillan, J. A., DeAngelis, C. D., Feigin, R. D., Warshaw, J. B. (1999). *Oski's pediatrics*. Philadelphia (PA): Lippincott Williams & Wilkins.
- Michie, S., Bron, F., Bobrow, M., Marteau, T. M. (1997). Nondirectiveness in genetic counselling: an empirical study'. *Am J Hum Genet* 60:40–47.
- Mill, J. S. (1978). *On liberty*. Indianapolis (IN): Hackett Publishing Company (originally published in 1859).
- Molewijk, A.C., Stiggelbout, A. M., Otten, W., Dupuis, H. M., Kievit. J. (1993). Implicit Normativity in Evidence-Based Medicine: A Plea for Integrated Empirical Ethics Research. *Health Care Analysis* 11:69-92.
- NHS Choices. Turner Syndrome. Retrieved on Dec 21 2013 from: <http://www.nhs.uk/conditions/turners-syndrome/Pages/Introduction.aspx>
- Nuffield Council on Bioethics (1993). *Genetic screening: ethical issues*. London: Nuffield Council on Bioethics.
- O'Neill, O. (2002). *Autonomy and trust in bioethics*. Cambridge: Cambridge University Press.
- Oduncu, F. 2002. The role of non-directiveness in genetic counselling. *Med Health Care Philos* 5:53 - 63.
- Pinsker, J. E. (2012). Turner Syndrome: Updating the Paradigm of Clinical Care. *Clin Endocrinol Metab* 97(6): E994 - E1003.
- Punales Morejon, D. (1996). Society's diseases [commentary]. *Hastings Cent Rep* 26(3): 21 - 22.
- Ranke, M. B., Saenger, P. (2001). Turner's syndrome. *Lancet* 358: 309 - 314.
- Rudolph, A.M. Hoffman, J. I. E. (1987). *Pediatrics*. Norwalk (CT): Appleton & Lange.
- Saenger, P. (2008). Turner Syndrome. In M. A. Sperling (ed.). *Pediatric Endocrinology*, 3<sup>rd</sup> edition. Philadelphia: Saunders/Elsevier 2008.
- Saenger, P., Wikland, K. A., Conway, G. S., Davenport, M., Gravholt, C. H., Hintz, R. & Silberbach, M. (2001). Recommendations for the diagnosis and management of Turner syndrome. *Journal of Clinical Endocrinology & Metabolism*, 86(7): 3061 - 3069.

- Somer, M., Mustonen, H., Norio, R. (1998). Evaluation of genetic counseling: recall of information, post-counseling reproduction, and attitude of the counselees. *Clin Genet* 34: 352 - 365.
- Sybert, V.P. McCauley, E. (2004). Medical progress: Turner's syndrome. *N Engl J Med* 351: 1227 - 1238.
- Turner Syndrome Society. Retrieved on Dec 21 2013 from: <http://www.turnersyndrome.org>
- Wertz, D. C., Fletcher, J.C. (1988). Attitude of genetic counselors: a multinational survey. *Am J Hum Genet* 42: 592 - 600.
- White, M. T. (1998). Decision-making through dialogue: reconfiguring autonomy in genetic counseling. *Theor Med Bioethics* 19:5–19.
- Wolff, G., Jung, C. (1995). Nondirectiveness and genetic counselling. *J Gen Couns* 4:3–25.
- Yarborough, M., Scott, J. A., Dixon, L. K. (1989). The role of beneficence in clinical genetics: non-directive counseling reconsidered, *Theor Med* 10:139–149.