

Communication with patients during the prenatal testing procedure: An explorative qualitative study

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Abstract

Objective: While generally two phases of prenatal genetic counseling are distinguished, i.e. pre- and post-test counseling, we revealed a third form of communication *during* the testing procedure. The content of this intermediate communication was explored.

Methods: A secondary analysis was performed on data obtained in another observational study, which was focussed on how indefinite testing results are clarified. Thirteen testing trajectories in which communication with parents took place during the testing procedure were further analysed.

Results: In the majority of cases the content of intermediate communication was similar to the content of pre-test counseling. In four cases the content was different, because the communication involved the parents in decision-making about a testing result, which was still being processed.

Conclusion: Communication in (prenatal) genetic testing is not always restricted to separate phases, but can be an ongoing process occurring parallel to, and sometimes even intertwined with, the testing process. The advocated model of shared decision-making might work better once it is determined if the decision concerns the area wherein the provider is the expert, or the patient.

Practice implications: Further research into the process of continuing decision-making could clarify how providers' and patients' responsibilities regarding the diagnostic process are distributed. Meanwhile, the possible occurrence of continuous decision-making should be mentioned in (prenatal) genetic counseling.

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1. Introduction

Our understanding of the genetic counseling process is far from ideal. Using the 'black box' metaphor, Biesecker and Peters recently stated that we know very little of the inside of the genetic counseling process [1]. There is little research documenting what counselors describe themselves as doing, or what they actually do, during the counseling process, nor is there a standard method of practice among prenatal clinic settings [2,3]. Overall, there is an inconsistency in defining the exact nature of counseling and its intended goals [4,5].

However, there is a consistent tendency to distinguish different phases of the genetic counseling process. In prenatal testing, two phases are generally characterized, i.e. pre-test counseling and post-test counseling. Pre-test counseling ideally includes information about the condition for which the testing is being offered, the characteristics of the test (including the chance of provoking a miscarriage), and the implications of possible test results. Post-test counseling consists of providing information about the diagnosis of a fetal abnormality, and providing emotional and decisional support regarding possible termination of pregnancy [6].

Another consistency in the literature is the idea of separating the genetic counseling interaction from establishing the medical genetics diagnosis [1]. Despite empirical studies showing clients often having difficulty distinguishing

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the counseling portion of the visit from the diagnostic or management portion [7], the idea that the counseling part takes place separately from the diagnostic part is rarely objected to. Even when an integrated approach to genetic testing is proposed, the different elements of the model are still presented as occurring separately in time [8].

Harper describes the genetic testing process as including three phases, i.e. (1) information, preparation and consent, (2) laboratory analysis, and (3) interpretation and support. In this model, the second phase starts when the first phase has ended, and the third phase starts when the second phase ends. Kessler, a well-known psychological expert in the field of genetic counseling, gives a similar description of the third phase of genetic testing: “The kind of genetic counseling session described here is one in which the data gathering and diagnostic activities have been *completed* and, following some unspecified period of time and/or rituals to mark off the session from what preceded it, the counsellors and counselees sit down together to discuss the diagnosis and its implication (including possible reproductive options) and attempt an integration [9].

Presented in terms of pre- and post-test counseling, the theoretical model of the prenatal testing process could thus be characterised as

- [A] Pre-test counseling: information, preparation, consent.
- [B] Diagnostic procedure (e.g. amniocentesis or chorionic villi sampling) + laboratory analysis, leading to a testing result.
- [C] Post-test counseling: interpretation, emotional and decisional support.

Most empirical research into prenatal genetic counseling will be focussed on either A or C, as these are the phases in the model in which physician–patient communication is expected to take place. However, in our observational study we also signalled some form of communication in phase B, i.e. *during* the testing procedure.

Because this had happened more than once we wondered if these observed kinds of communication between pre- and post-test counseling were just some rare exceptions to a model which was basically right, or if this model was perhaps too limited to reflect the day-to-day reality about prenatal genetic counseling. If the latter were the case, then the general distinction between pre- and post-test counseling, separated by the diagnostic procedure and lab analysis, might be an obstacle in our search for understanding the process of prenatal genetic counseling, and perhaps even of genetic counseling as such.

For this reason we decided to find out more about this communication with parents during the prenatal testing procedure. Was the communication in these contacts similar to what is normally discussed in pre- or post-test counseling, or was something else happening? And if so, how could this communication during the testing process be understood within the above-mentioned A-B-C model of prenatal

testing? Our research question was: “What was the content of the provider–patient communication in phase B of the prenatal testing process?”

More generally, we also examined if more insight into this communication during the testing process would possibly add to our *overall* understanding of the process of prenatal genetic counseling.

2. Methods

The above-mentioned forms of communication were accidentally found in a larger observational study about which we have reported elsewhere [10]. For the current study, we performed a secondary qualitative analysis on data we had already obtained in this larger study. Because we had not included the provider–patient communication in the original study design, we could only examine this data in an explorative way.

2.1. Data collection

The larger observational study took place in the periods April–June 2001, and July–September 2002. Observations were then focussed on results of which it was still unclear whether they would turn out to be normal or aberrant. These indefinite results, which we referred to as ‘grey’ results, are quite common in prenatal diagnosis (see Box 1). The observations were used to examine what professionals do to clarify grey results, i.e. to make them black or white.

In this larger study, a procedure was agreed with the respective professionals to ensure that the researcher could be present at the department for observation at key occasions. The actual observations concerned the interdisciplinary consultations between the professionals during the process of clarifying the grey result. Consultations between technician and cytogeneticist, between cytogeneticist and clinical geneticist, and between clinical geneticist and gynaecologist were observed. The weekly interdisciplinary meeting of the entire professional team involved in prenatal diagnosis, and the weekly technicians meeting, if relevant, were also observed.

In terms of the theoretical model of prenatal testing, as mentioned in the introduction, our observations in this larger study had been focussed on phase B. We had therefore not included observations of any communication with parents in our original study design, as we had assumed that such communication would only take place either in phase A or C. So by the time we had found out that such communication did take place in phase B, we could only make use of indirect observations of this communication, i.e. through what the professionals had reported about their communication with parents in phase B. The clinical geneticists, who were the professionals communicating with the parents, had reported about these contacts either in the interdisciplinary meetings, or directly to the researcher. However, all these indirect

Box 1. Grey results in prenatal chromosome diagnosis

The most commonly found chromosome abnormality in prenatal diagnosis is trisomy 21, which means that every cell contains three instead of two chromosomes 21. A trisomy 21 found in prenatal diagnosis is a definite testing results. Apart from definite testing results, prenatal diagnosis can also lead to indefinite testing results.

Definite testing results

Trisomy 21 leads to a combination of physical and mental disabilities (varying in severity), which is known as **Down's syndrome**.

Other definite chromosome aberrations have phenotypical consequences, which are generally considered as **more serious than Down's syndrome**. The most commonly known aberrations are trisomy 13 (in which three instead of two chromosomes 13 are present), trisomy 18 (three instead of two chromosomes 18) and triploidy (three chromosomes instead of two for all chromosomes). All these chromosomal aberrations result in severe physical and mental disabilities.

Another group of definite chromosome aberrations have phenotypical consequences, which are generally considered as **less serious than Down's syndrome**. Most common in this category are the sex chromosomal aberrations, in which case there is something wrong with the X- or Y-chromosomes. Among these, Turner's syndrome and Klinefelter syndrome are quite familiar. With Turner's syndrome (45,X) we find one single X-chromosome instead of two sex chromosomes; with Klinefelter syndrome (47,XXY) there is one X-chromosome too many. Most phenotypical problems in Turner's and Klinefelter's syndrome relate to infertility and/or secondary gender characteristics. In case of a 45,X or 47,XXY that appears in mosaicism these phenotypical problems are less serious (see the next section).

Indefinite (grey) testing results

In addition to the various types of definitive aberrant results, there are also chromosomal aberrations of which it still remains unclear if they will have phenotypical consequences. Among these indefinite, or grey, testing results, a distinction can be made between mosaicism and structural aberrations.

In **mosaicism** an individual shows two (or more) genetically different cell types. Forms of mosaicism have been observed of e.g. normal cells and cells with trisomy 21, or of normal cells and cells missing an X-chromosome, but mosaicism may appear in all kinds of variations.

Mosaicism is a relatively common phenomenon in chorionic villi sampling. As in chorionic villi sampling placenta material is examined and no fetal material, the observed mosaicism may be restricted to the placenta, but not manifest itself in the fetus. In that case one speaks of confined placental mosaicism. In chorionic villi sampling two different kinds of methods can be used to look at two types of cells of the placenta. When mosaicism is observed in the short-term culture (STC) of the chorionic villi sampling, additional testing can take place through a long-term culture (LTC). When mosaicism is *not* observed in the long-term culture, there is an increased chance that mosaicism is limited to the placenta, in which case it does *not* lead to phenotypical abnormalities.

In addition to mosaicism confined to the placenta, there may also be other explanations for mosaicism in laboratory material that cannot be found in the fetus. For instance because genetically deviating cells have developed in the cell culture (culture artefact) or because not only fetal cells but also cell material of the mother has been examined (maternal contamination). Culture artefacts and maternal contamination can both occur in chorionic villi sampling as well as in amniocentesis. In all these cases the detected mosaicism does not have phenotypical consequences.

In a **structural aberration, e.g. a translocation**, the problem concerns the form (structure) of the chromosomes. There are two kinds of structural aberrations. With an unbalanced structural aberration, the form of one or more of the chromosomes has been altered in such a way that the total amount of genetic material has also changed. In a balanced structural aberration, the total amount of (chromosomal) material has remained the same. Whereas an unbalanced aberration always leads to phenotypical aberrations, this is commonly not the case with a balanced aberration. To determine if the chromosomal aberration would lead to phenotypical aberrations, the chromosomes of the parents are analysed, as this may indicate whether the detected change of the chromosomes occurs within the family or is a new alteration. When one of the parents is carrier of the same chromosome anomaly, it is not to be expected that there will be phenotypical aberrations; for the parent with the same structural aberration is of normal health. But when it concerns a new aberration, it cannot be excluded that the structural aberration will have phenotypical consequences.

reports had been included in the field notes, and were therefore available in the observation protocol.

Apart from these direct and indirect observation data, the patient files, containing laboratory forms, the result letter and all other correspondence with medical specialists, were also collected. Finally, the minutes of the weekly interdisciplinary meetings were examined.

2.2. Data analysis

All research material was structured by ‘testing trajectory’, i.e. the series of professional actions through which a grey testing result was clarified. To this end, all fragments from the observation protocol relating to the same testing trajectory were put in chronological order. Kwalitan, a software programme specially developed for analysing qualitative data, was used for this purpose [11]. The next step was working out the reconstructed trajectories into comprehensive case reports, always taking the primary research data into account. These case reports were checked by a cytogeneticist (LK) and a clinical geneticist (NL) of the department, to detect factual irregularities, but also as a form of ‘member checking’, i.e. as a tool to guarantee the validity of qualitative research [12,13].

In thirteen of the observed grey testing trajectories some form of communication with parents had been taken place during the testing process. These 13 grey trajectories were further analysed for this study. These trajectories were considered as case studies, which were both studied individually (individual case analysis) as well as in relation to each other (cross-case analysis) [14]. The qualitative analysis was directed by the research question. By carefully examining the reconstructions of the testing trajectories it was determined how the communication during the testing process related to the process of clarifying the grey result. This qualitative analysis was performed by the first author, and checked by NL and LK.

3. Results

In all 13 analysed grey testing routes the *timing* of communication with parents differed from the phase in which pre- or post-test counseling normally occurs, as it took place during the testing procedure. Due to this different timing we refer to this as ‘intermediate communication’. In all cases this intermediate communication procedure took place because the testing trajectory had resulted in an indefinite result for which clarification was needed.

However, the *content* of the intermediate communication with regard to the clarification of the indefinite testing result differed among the 13 testing trajectories. Whereas in the majority of cases (9/13) the intermediate communication was about medical and/or technical matters only, in four cases more personal matters were

discussed as well. Table 1 gives an overview of all thirteen observed cases of intermediate communication during the testing procedure.

3.1. Communicating medical/technical matters only

In two cases the parents were informed that the testing route would take a little longer because a second laboratory analysis was necessary to clarify the indefinite result. In the other seven cases the parents were not only informed about the delay of the laboratory procedure, but they were also informed about, and asked consent for a second diagnostic procedure, either amniocentesis ($n = 3$), ultrasound examination of the fetus ($n = 1$) or examination of the parents’ blood ($n = 3$).

3.2. Communicating a mix of medical/technical and personal matters

In four cases, apart from medical/technical details, communication was also about the parents’ personal evaluation of the testing result. Two of these cases were quite similar. The indefinite result found in trajectory 15 was a mosaic 45,X (mosaic Turner, see Box 1), and in trajectory 19 it was a mosaic 46,XY/45,X. When the parents were informed about the additional testing necessary to clarify this result, the parents’ personal considerations on this situation were also discussed.

The clinical geneticist who had contacted the parents of trajectory 15 reported in the weekly interdisciplinary meeting that “*they were merely very pleased that their child did not have Down’s syndrome; a girl with Turner’s syndrome would be quite acceptable to them.*”

The parents had let the clinical geneticist know that, while they might have terminated their pregnancy in case trisomy 21 (Down’s syndrome) would have been detected, they would not do this for mosaic 45,X. This intermediate communication influenced the proceedings of the testing trajectory. Because the parents would not terminate this pregnancy, it was decided not to do amniocentesis to clarify the indefinite mosaic.

The clinical geneticist who had contacted the parents of trajectory 19 had explained to the parents there was a 10% chance there would be something wrong with the genital formation. In the weekly interdisciplinary meeting the clinical geneticist reported: “*Well, this was entirely acceptable to them (...) they were more than willing to take their chances in this matter.*” Consequently, because these parents would continue this pregnancy anyway, no matter if the amniocentesis would detect the mosaicism or not, it was decided not to do amniocentesis.

The indefinite result found in trajectory 25 was a mosaic 47,XXX/45,X. Because of additional testing the parents needed to wait a little longer for a definitive testing result. They were also informed about the possible impact of the indefinite mosaic, i.e. the possibility that the ovaries might

Table 1
Cases of observed intermediate communication during testing procedure ($n = 13$)

Content of communication	Indefinite (grey) testing result	Indication	Initial procedure	Possible additional testing	Content of in-between communication: summary
Medical/technical matters (n = 9)	(20) ^a Mosaic ^b trisomy 21	Advanced maternal age	CVS ^c (STC ^d)	LTC ^e	The parents were informed that their testing result would take a little longer because of the LTC
	(18) Mosaic trisomy 18	Advanced maternal age	CVS (STC)	LTC	Idem
	(29) Mosaic trisomy 18	Advanced maternal age	CVS (STC)	Amniocentesis (not enough material for LTC)	The parents were prepared and asked consent for amniocentesis
	(27) Mosaic trisomy 2, trisomy 6, trisomy 7	Risk of DNA abnormality ^f	CVS (STC)	Amniocentesis	Idem
	(38) Mosaic trisomy 18 and trisomy 7	Advanced maternal age	CVS (STC)	Amniocentesis (not enough material for LTC)	Idem
	(17) Mosaic 46,XX/46,XY	Advanced maternal age	Amniocentesis	Ultrasound examination of the fetus	The parents were prepared and asked consent for ultrasound examination of the fetus
	(23) Balanced translocation ^g	Advanced maternal age	Amniocentesis	Parental chromosome analysis	The parents were prepared and asked consent for examination of the parents' blood
	(32) Balanced translocation	Previous child with chr. abn.	CVS	Parental chromosome analysis	Idem
	(34) Balanced translocation	Advanced maternal age	Amniocentesis	Parental chromosome analysis	Idem
Mix of medical/technical and personal matters (n = 4)	(15)* Mosaic 45,X	ICSI ^h and NT ⁱ result	CVS (STC)	LTC, and amniocentesis if the LTC would show the mosaic 45,X/46,XX	The parents communicated that the uncertainty of this grey result was acceptable to them, now they knew their child did not have Down's syndrome. They would continue the pregnancy anyhow, independent of the testing result of the LTC, and did not want amniocentesis
	(19)* Mosaic 46,XY/45,X	Risk for DNA abnormality	CVS (STC)	Amniocentesis (not enough material for LTC)	The parents communicated that the uncertainty of this grey result was acceptable to them, now they knew their child did not have the DNA abnormality. They would continue the pregnancy, and did not want amniocentesis
	(25)* Mosaic 47,XXX/45,X	Advanced maternal age and triple test ^j result	Amniocentesis	i-FISH ^k	The parents communicated that the uncertainty of this grey result was not acceptable to them. They did not wait for the i-FISH result, and decided to terminate the pregnancy
	(24) Fear of mosaicism	Advanced maternal age	Amniocentesis	i-FISH and/or amniocentesis	The parents communicated that for them there were no specific individual reasons to accept or not accept the uncertainty of this grey result. i-FISH was done to clarify this grey result

^a () = Number of observed trajectory. The trajectories marked with * are also described in detail in Zwieten et al. [10].

^b See Box 1 for explanation of mosaicism.

^c CVS = chorionic villi sampling.

^d STC = short-term culture; see Box 1 under mosaicism for explanation.

^e LTC = long-term culture; see Box 1 under mosaicism for explanation.

^f When prenatal diagnosis is performed because of an increased risk for a specific DNA abnormality, it is standard procedure – after the patient's consent – to analyse the chromosomes as well.

^g See Box 1 for explanation of balanced translocation.

^h Intra cytoplasmic sperm injection (ICSI) is a form of IVF in which a sperm cell is injected directly into the egg cell. An ICSI procedure is presumed to lead to an increased risk for chromosomal abnormalities [29].

ⁱ Nuchal translucency (NT) measuring is a form of prenatal screening in which the thickness of the nuchal translucency is measured through an ultrasound scan. A thickened nuchal translucency indicates an increased risk for Down's syndrome and other chromosomal abnormalities.

^j The tripletest is a measurement of three substances in the blood of the pregnant woman and results, combined with the age of the pregnant woman, in a calculated risk for Down's syndrome.

^k In interphase fluorescent in situ hybridisation (i-FISH) more amniotic fluid cells can be analysed than with the conventional method of karyotyping, for which only dividing cells are used. i-FISH can be performed on cell material which is already available from amniocentesis, so there is no need for a second invasive procedure.

hardly develop, implying a possible infertility. Additionally, they were warned not to look for info on the Internet on their own, because the symptoms mentioned there were far too serious. However, a few days later these parents told their gynaecologist: “*We don’t dare to take the risk after all.*” According to the clinical geneticist: “*Among others, because all information on the net about Turner had really scared them.*” The parents’ personal considerations had made them decide to terminate the pregnancy, no matter the result of the additional test. Consequently, the result of this test was not communicated to the parents anymore.

The grey result in trajectory 24 was not so much an actual detected aberration, but more the *suspicion* of mosaicism. Due to technical reasons the geneticists did not feel confident to give a definitive testing result, because they first wanted to exclude the possibility of mosaicism. The two options to do this were discussed with the parents, seemingly to find out how the parents would wish the testing procedure to continue. However, these parents did not seem to have any strong personal considerations regarding this matter. Consequently, it was decided to choose the option which was most reasonable from the professionals’ technical perspective.

3.3. Results in terms of the A-B-C model

The intermediate communication in the nine cases in which only medical/technical matters were discussed can be understood as a repeated form of pre-test counseling. In terms of the A-B-C sequence this is what happened in these cases:

- [A] Pre-test counseling.
- [B] Diagnostic procedure + laboratory analysis, leading to an *indefinite* result.
- [I] Intermediate communication = second pre-test counseling [A2].
- [B2] Second diagnostic procedure and/or laboratory analysis, leading to a definitive testing result.
- [C] Post-test counseling.

In the other four cases the content was very different to the content of pre- or post-test counseling as the intermediate communication (I) seemed to be *intertwined* with the proceeding of the diagnostic procedure and laboratory analysis (B2), as well as with post-test counseling (C). Therefore, in terms of the A-B-C model, this is what happened in these four cases:

- [A] Pre-test counseling.
- [B] Diagnostic procedure + laboratory analysis, leading to an indefinite testing result.
- [I + B2 + C] Intermediate communication, intertwined with the proceeding of the diagnostic procedure + laboratory analysis, and with post-test counseling.

3.4. Decision-making during testing process

The intertwined intermediate communication with parents in the last four cases was directing the testing process, because through this communication the goal of prenatal diagnosis was specified. But why was the goal of prenatal diagnosis specified *during* the testing process? Due to technical characteristics, the goal of prenatal cytogenetic diagnosis is by definition not always very specific. Since full karyotype analysis is the ‘gold standard’, generally *all* chromosomes are examined in order to find a single chromosome abnormality. This means that chromosome analysis might lead to the detection of *any* kind of chromosome abnormality, not only the one for which the test is actually performed because of an existing high risk. In these cases, the detected aberration is an unexpected finding for the patient [15]. In the four cases which have been described in detail, the chromosome abnormality found was grey (see Box 1), but unexpected for the parents too. Because the detection of a so-called unexpected finding is a well known phenomenon in prenatal diagnosis, it is advised to inform parents about this possibility in pre-test counseling [16–19]. However, in daily practice this is rarely done [20].

We do not know if the parents in our observations were actually informed beforehand about these possibilities or not, but we do know from the observations that the parents were informed about the unexpected, grey result while the testing procedure was already going on. Whereas in nine cases the parents were indeed informed that this grey result needed to be further clarified, in the other four cases the communication with the parents dealt with the question if and how the grey result should be clarified. As such, the intermediate communication in these cases led to a process of decision-making, which was not located either in pre- or post-test counseling. Instead, this process of the parents’ decision-making took place *during* the testing process.

4. Discussion and conclusion

4.1. Discussion

The content of the provider–patient communication during the prenatal testing process was similar to the content of pre-test counseling in the majority of cases. In four cases the content was different, because the parents were involved in decision-making about a testing results, which was still being processed.

This study presents a secondary analysis on data that was already available through another study. Although the small number of cases limit the external validity of our study [21,22], our results may still help to better understand the process of genetic counseling in general.

Firstly, communication in prenatal genetic testing evidently does not only happen in separate phases before or after the test, but also during the testing process. The fact

that parents sometimes have to make decisions during the testing process has been signalled before [23]. Communication, including the parents' process of decision-making, may therefore be thought of as an ongoing process, occurring parallel to, and sometimes even intertwined with, the testing process.

Secondly, the cases in which the parents took part in deciding how to proceed with the testing process can be interpreted as examples of shared decision-making, a model, which is recently being promoted to apply in genetic counseling [24,25]. It is already acknowledged that shared decision-making may also be applicable to negotiations about diagnostic pathways [24]. However, based on the results of this study, we are not convinced that involving patients in the diagnostic pathway of their own testing results should be considered a goal in itself. On the contrary, we endorse the viewpoint that in cases of shared decision-making clients should not feel abandoned to make important decisions without sufficient support, when counsellors, for example, withdraw from any involvement [24]. As professionals in genetic counseling are used to thinking of communication with their clients in terms of a consumer model, rather than the paternalistic model as is more common in other medical settings [26], this pitfall seems realistic. Therefore, when applying the model of shared decision-making in genetic testing settings, it could be kept in mind that the patient leads in areas where he is the expert, and the doctor leads in his domain of expertise [27]. Shared decision-making might work better once it is determined if the decision to be made concerns the area wherein the provider is the expert, or the patient. Doing this might prevent the parents to feel abandoned in an area like the diagnostic pathway, which is, due to the complex technical character, primarily the doctor's domain of expertise.

4.2. Conclusion

This explorative study illustrates that parents could also be involved in decision-making while the testing result is still being processed. Due to some particular features of prenatal testing, e.g. uncertainty and the highly complex character of the testing procedures, it remains to be determined if parents experience this as a gain or as a loss. In evaluating the increasingly advocated communication model of shared decision-making, parents' appreciation of continuing decision-making should be weighed as well.

4.3. Practice implications

The occurrence of continuing decision-making shown in this study should be further examined. Particular attention should be focussed on how responsibilities between providers and patients are distributed in this process, for example along the lines suggested by Salmon and Young in their recent paper [28]. Meanwhile, counseling and

education about (prenatal) genetic testing might emphasise that decision-making for parents involved in prenatal testing may exceed pre- and post-test counseling.

In this paper, we mainly focussed on the professionals' part of the communication process. However, an important step towards opening the 'black box' of genetic counseling might be to examine the clients' experiences with indefinite results of genetic testing. Further study of the communication following indefinite testing results and of other intermediate communication will definitely add to the overall understanding of the genetic counseling process.

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