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Genetic counseling in veterinary medicine: towards an evidence-based definition for the small animal practice

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Abstract

Background In human medicine, questions regarding heritable disorders are dealt with by clinical geneticists and genetic counselors and both the field, their roles and the tools they use are well-defined. Even though the prevalence of diseases is far higher and scientific literature agrees on expectations towards an increased importance, this does not seem to be the case in veterinary medicine. While we hypothesize that there will be an overlap, some characteristics uniquely linked to veterinary medicine might not be covered.

Methods To investigate this in-depth and in an attempt to define the field, we compared the internationally accepted definitions and its subparts on genetic counseling in human medicine with what is found in veterinary literature and what was seen in cats and dogs presented at our dedicated small animals clinical genetics/genetic counseling clinic. The results were used in a stepwise analysis that lead to a set of three potential definitions (i.e. on what genetic counseling is, who provides it and which tools are used) that fullfill four criteria (i.e. definitions have to be clear/self-explanatory, minimally sufficient, complete and valid).

Results The short version of the definition of genetic counseling in veterinary medicine is: "Genetic counseling is the process of helping animal owners and breeders understand – and adapt to – the medical, psychological, familial implications of genetic contributions to disease." Genetic counseling in small animal practice is currently provided by veterinarians and the tools that are used, can be divided in five categories. The signalment of the patients revealed that both cats (30%) and dogs (70%) and various breeds, the two sexes (37% males, 63% females) and all age categories (puppy/kitten—senior) were represented. Furthermore, 73% of the patients were referred by or needed to be referred to other disciplines.

Conclusion These definitions are derived from human and veterinary literature, and an evaluation based on patient data has demonstrated that these definitions meet all the criteria of a correct definition (i.e. clear, minimally sufficient, complete and valid). With these definitions and case descriptions, our aim is to contribute to the formal foundation of genetic counseling in veterinary medicine.

Keywords Clinical genetics, Genetic counseling, Small animal medicine, Veterinary medicine

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Introduction

While cats and dogs are part of a lot of households and often seen as man's best friends, various ethical questions have been raised and concerns expressed regarding their welfare. Documentaries (e.g. 'Pedigree Dogs Exposed') have led to a shockwave among the general public and the number of bans imposed on certain cat and dog breeds in various countries is increasing [1]. The origin of these claims and legal bans are linked to the observations that both dogs and cats suffer from a variety of genetic diseases, like canine brachycephalic obstructive airway syndrome and osteochondrodysplasia in the Scottish Fold, at high frequencies, with some of them associated with breed standards [2–5].

These observations are not new and linked to the population history and breeding practices. Practices such as the popular sire effect, where a male animal with desirable attributes has a disproportionately large progeny, or line- and inbreeding and population bottlenecks were – and still are – common in dog breeding. These practices are not harmless and have increased the level of homozygosity for detrimental alleles, which leads to an increased prevalence of inherited disorders, and decreased genetic diversity [6, 7]. Similar observations have been made in cats, albeit their genetic diversity is often less reduced [8].

To reduce the number and prevalence of inherited disorders, various strategies and tools can be used. They often involve (a combination of) screening programs, genetic testing, and selection methods (e.g. estimated breeding values). While they can be used in any breeding program, their proper use is especially critical in rare breeds with a small or decreasing population size, and for breeds predisposed to a high number of inherited disorders [9]. Eventhough there are of course some notable examples where the prevalence of diseases was successfully reduced (e.g. the prevalence of patellar luxation in the Dutch Kooikerhondje has decreased from 28 to 19% and the prevalence of hip dysplasia in a population of Labrador Retriever assistance dogs from 41 to 0% or even the reduction in the occurrence of myxomatous mitral valve disease in Cavalier King Charles Spaniels), several reports seem to be alarming rather than comforting when it comes to genetic diseases in companion animals [6, 7, 10].

In human medicine, questions regarding heritable disorders are dealt with by clinical geneticists and genetic counselors. Despite local differences linked to country-specific characteristics of the healthcare system, legal restrictions, and cultural factors, the profession shares many similarities globally [11]. With projects like the Human Genome Project, an international scientific effort that aimed to map and sequence the entire human genome [12], and the subsequent improvements in technology and understanding of genetic diseases, the demand for genetic counseling in medicine has exponentially risen over the years. Since the foundation of The National Society of Genetic Counselors (NSGC) in 1979, an organization that "advances the various roles of genetic counselors in health care by fostering education, research, and public policy to ensure the availability of quality genetic services", the profession has expanded globally, and in early 2018, they estimated that there were nearly 7,000 genetic counselors in over 28 countries [11]. In 2003, genetic counseling was defined by the NSGC. Furthermore, the various actors are well known and (international) training programs have been set up. Surprisingly, in veterinary medicine, this does not seem to be the case, eventhough the prevalence of diseases is far higher and scientific literature agrees on expectations towards an increased importance [13].

To investigate this in-depth, we compared the internationally accepted definition and its subparts on genetic counseling in human medicine with what is found in veterinary literature and what was seen in cats and dogs presented at our dedicated small animals clinical genetics/genetic counseling clinic.

The main goal was to provide answers to the following research questions:

- Question 1: what is genetic counseling?
- Question 2: who provides genetic counseling?
- Question 3: what tools are used in genetic counseling?

Additional aims were to characterize cats and dogs patient characteristics (i.e. signalment) and to position genetic counseling relative to other disciplines in veterinary medicine.

We intially expected that the definition for what genetic counseling is in human medicine will overlap largely, given our observations from the dogs and cats in the clinic. As the canine and feline population have several characteristics which are relatively unique compared to humans, we expected some items to be missing however. Taking the sparsity of literature in account, we expected that the veterinary literature would not sufficiently address these questions.

Materials and methods

Research questions

A stepwise analysis was used that lead to a set of three potential definitions (i.e. on what genetic counseling is, who provides it and which tools are used) that fullfill four criteria (i.e. definitions that are clear/self-explanatory, minimally sufficient, complete and valid) [14].

Data sources and search strategy

To answer these questions, three sources were used:

The first (literature-based) source consists of 1/ the combination of definitions developed by the NSGC [15] and 2/ the reference handbook: 'A guide to genetic counseling' [16]. Two veterinary genetic counselors (L.A. and B.J.G.B) retrieved the definitions. An experienced and NSGC board-certified genetic counselor with > 10 years of experience (V.S.) checked whether the retrieved definitions appropriately described the situation in human medicine.

The second source was literature in PubMed. Two separate PubMed searches were performed with the following terms: 1/ "Genetic counseling" AND "medicine" and 2/ "Genetic counseling" AND "veterinary medicine". The results from these two searches were used to allow a cross species comparison on the amount of scientific literature available, while the results for the second search (i.e. the one with "veterinary medicine") were used for the subsequent in-depth analysis. The strategy is detailed in a PRISMA flow diagram (Fig. 1) [17]. Exclusion criteria for the latter were articles solely focusing on humans. Articles that were not accessible, even after requests, were also excluded. Articles were not limited to geographical location and included clinical trials, reviews and systematic reviews. An overview is provided in Supplementary File S1.

The last source consists of the cats and dogs presented to our dedicated small animals clinical genetics/genetic counseling service (i.e. the advice center of clinical genetics for companion animals at the Faculty of Veterinary Medicine, Ghent University). The clinical data of each cat

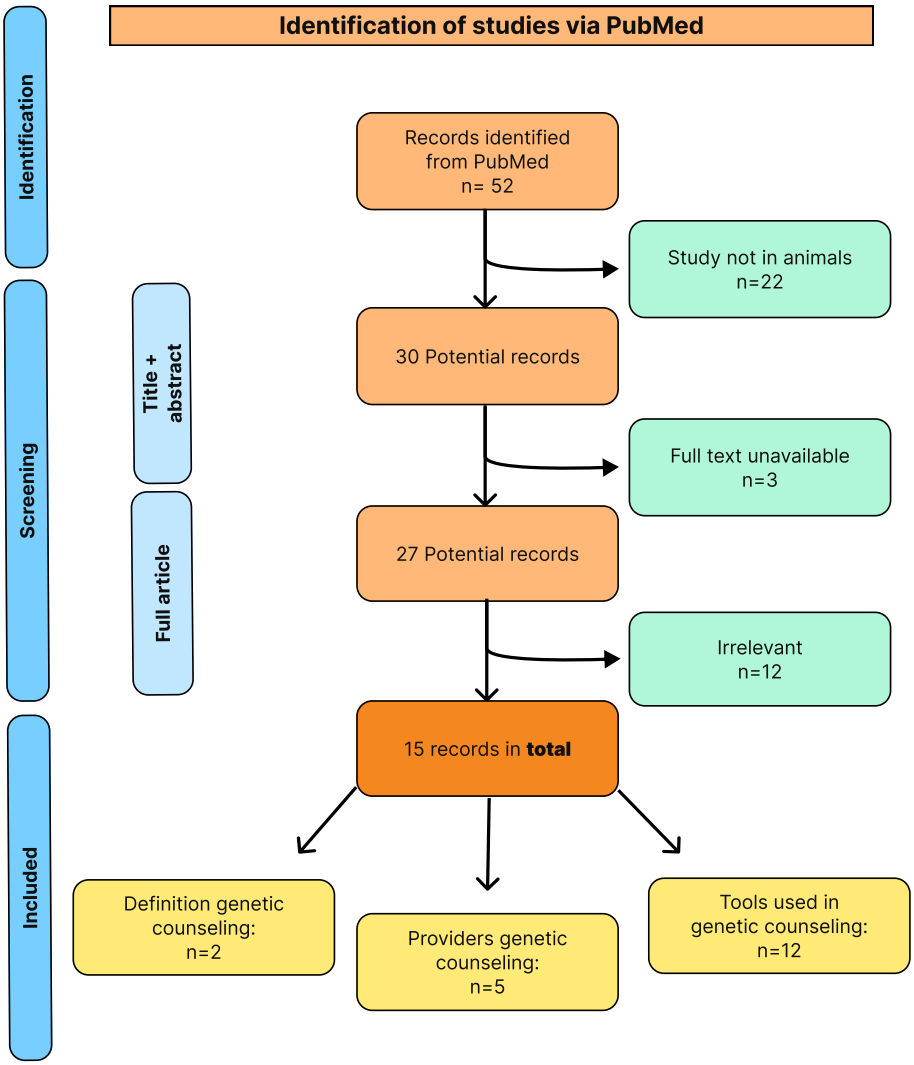

Fig. 1 A PRISMA flow diagram providing an overview of how the records from PubMed were handled

and dog was retrospectively reviewed (Supplementary File S2).

Analysis

Adhering to the methodology used in another study dealing with defining a procedure, the goal was to obtain definitions that are clear/self-explanatory, minimally sufficient, complete and valid [14]. These evaluations were performed independently by two veterinary genetic counselors (L.A. and B.G.J.B) and results were compared. Practically, this translates to going over the data and answering the following questions:

- Clear/self-explanatory: is everything clearly phrased, i.e. is everything in the definition phrased in a manner that requires little explanation beyond an understanding of the individual words from which it is constructed? (yes/no)
- Minimally sufficient: are there redundant items? (yes/no)
- Complete: are all consultation characteristics completely described by the literature-based definitions? (yes/no). An assessment of the completeness thus starts from the feline/canine patients' point of view and evaluates whether the definitions adequately describe their situation.
- Item validity: does every item mentioned in the literature-based definitions have a match in the consultation characteristics? (yes/no per part). An assessment of the validity thus starts from the literature's point of view.

A stepwise analysis was conducted (Fig. 2). In step one, (individual parts of the) literature-based definitions were evaluated on their clarity/self-explanatoryness and potential redundancy. In step two, the literature-based

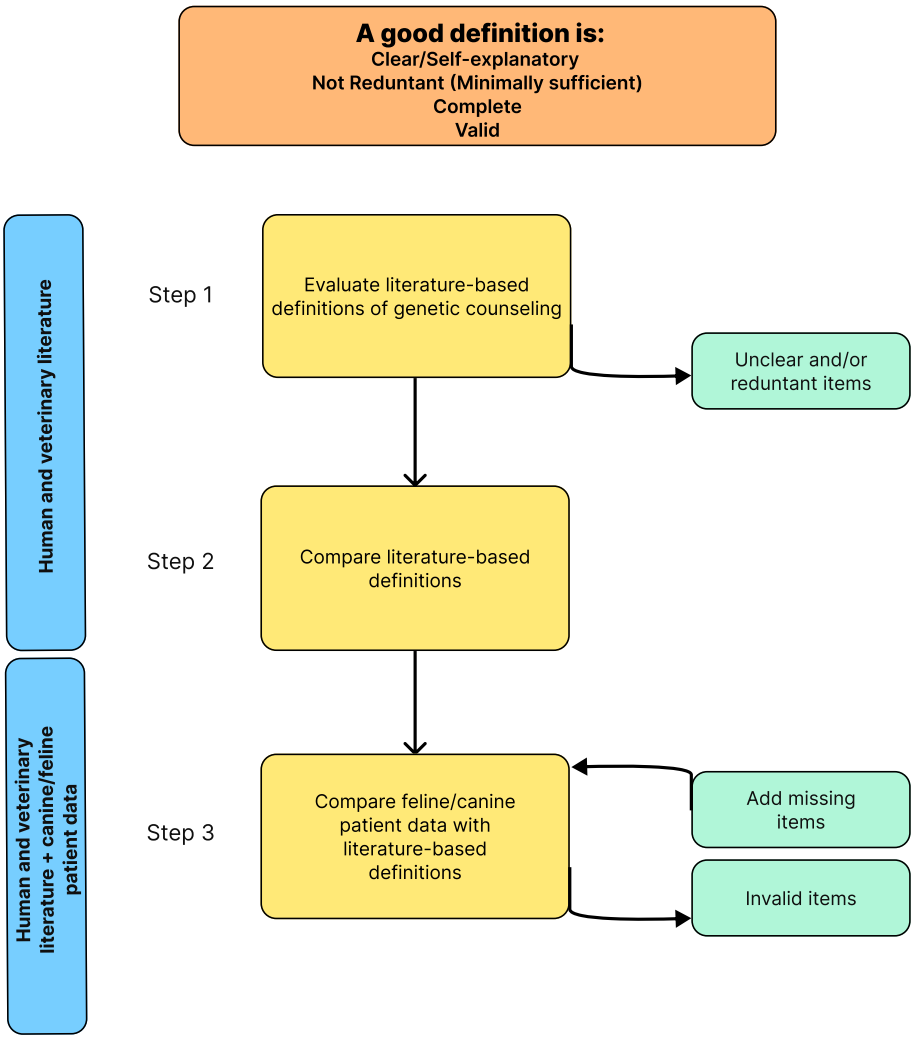

Fig. 2 A stepwise analysis was used to obtain a definition that is clear/self-explanatory, non redundant/minimally sufficient, complete and valid

definitions of genetic counseling in medicine and veterinary medicine (source one and two) were compared to evaluate whether they matched or not. In step three, to assess the completeness and the validity, the feline/canine patient data (=source three) was compared with the definitions retrieved from source one and two, respectively. All steps in the analysis were conducted independently and in parallel by two authors (L.A. and B.J.G.B) to minimize the effect of personal interpretation. If there was no agreement, this was noted and subsequently discussed until consensus was reached.

Results

Literature search results

In medical literature, a total of 10,231 records were retrieved, with a peak of 1,053 articles in 2021 and all articles being published between 1953 and 2023. In veterinary medicine, the largest number of articles was published in 2017, and all articles were published between 1986 and 2023 (Fig. 3). An overview of the literature-based definitions for genetic counseling in medicine is provided in Table 1.

The veterinary literature search yielded 52 potentially relevant records (Supplementary File S1). Out of 52 potentially relevant citations, 22 articles were excluded because they solely described disorders in human medicine. From the remaining 30 articles, 4 were initially inaccessible, of which 3 remained unavailable even after contacting the authors. As such, a total of 27 articles remained. From these 27 articles, several did not provide an answer to any of the three main questions, so finally, 15 articles were potentially useful to define genetic counseling in veterinary medicine (Table 1). Two articles contain information relating to the definition of genetic counseling, 5 articles describe who provides it and 12 articles refer to tools that can be used in genetic counseling in veterinary medicine (Fig. 1).

Step 1: evaluation of clarity and redundancy

An overview of the literature-based results that were used to evaluate clarity and redundancy are provided in Table 1. For the results in medicine, both evaluators unanimously agreed that the various definitions (and their subparts) were clear and did not contain redundant information on any of the three questions.

For veterinary medicine, on only one of the three questions (the first question on what genetic counseling is), both evaluators unanimously agreed that the information derived from literature was clear and did not contain redundant information. On the second question ("who provides genetic counseling"), both evaluators agreed that it was unclear what was meant by "professionals". In the third question i.e. the tool-based question, "non-DNA-based genetic counseling" was also considered to be an unclear component by one evaluator and when the item was discussed, it turned out that, depending on the evaluator, a different meaning was given to it. As such, this term is by definition unclear. The remaining items were considered to be clear and non-redundant and were used for the subsequent analyses.

Step 2: comparison of genetic counseling in medicine versus veterinary literature

A comparison of the remaining items in Table 1 was subsequently performed. For question 1, veteri-

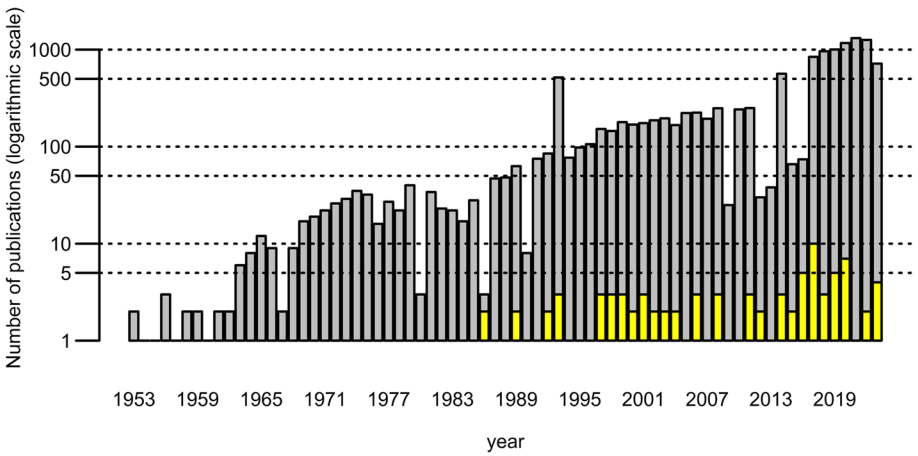

Fig. 3 An overview of the number of publications per year that matched the search terms for genetic counseling in human medicine (black bars) and genetic counseling in veterinary medicine (yellow bars), respectively. The y-axis is on a logarithmic scale due to the pronounced count difference

Table 1 PubMed search

Questions	Source 1	Source 2
What is genetic counseling?	Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following: - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence - Education about inheritance, testing, management, prevention, resources and research - Counseling to promote informed choices and adaptation to the risk or condition [15, 16]	Prevention (2/2) [18, 19]
Who provides genetic counseling?	Genetic counselors consist of a variety of specialists from different fields and are not exclusively medical doctors [16]	Veterinarians (3/5) [13, 20, 21] Genetics researchers (1/5) [22] *Professionals* (1/5) [21] Veterinary genetic counselors (1/5) [23]
What tools are used in genetic counseling?	Tools: - essential for information gathering: e.g. family history (pedigrees) - used for establishing/verifying diagnosis: this includes DNA-testing, among other tests - related to risk assessment, i.e. they are used to calculate future reproductive or personal health risk - related to information giving, i.e. to help the patient understand the process and the implications of the findings for the affected person and family - related to psychological ounselling and support, e.g. to help the patient deal with emotional responses after giving distressing data [16]	Pedigrees of animals (1/12) [24] DNA-tests (9/12) [13, 18, 19, 23, 25–29] Heritability for risk estimation (2/12) [30, 31] *Non-DNA-based genetic counseling* (1/12) [19] Effective population size (1/12) [18]

Overview of the answers to the main research questions from the human medicine (= source 1) and veterinary literature (= source 2) point-of-view. Items considered to be unclear are in italic. For source 2, counts (between brackets) are provided for the number of times a specific item was mentioned, relative to the total number of references that provided an answer on specific question

nary literature contains only one item, while the definition in medicine can be divided in four subparts. Prevention, the only item from veterinary literature, was deemed to be part both of the "education" and "counseling" part of the definition in medicine. On the other hand, all items found in medicine were deemed to encompass more than "prevention" alone.

For question 2, which aims to provide an overview of the care providers, two items matched: medical doctors in human medicine and veterinarians in veterinary medicine are each other's counterpart, and genetic counseling were also mentioned in both human and veterinary sources. Genetics researchers were however mentioned only in veterinary literature and were thus considered to be a missing item in human medicine.

For question 3, all items except "effective population size" could be matched with the items from medicine, while "information giving" and "pschycological counseling" were not mentioned in veterinary literature. Nevertheless, the tools description found in medicine seemed to encompass more than the tools specifically mentioned in veterinary literature.

Step 3: evaluating completeness and validity: a comparison of the cats and dogs data with the literature

A total of 30 consultations were conducted between 2022 and 2023. An overview of the completeness per question is provided in Table 2. Per-cat/dog data on the validity of each item and the completeness are provided in Supplementary File S2 and S3, respectively. For the question on what genetic counseling entails, the medicine-based description of genetic counseling was sufficiently complete to describe 80% (24/30) of the cats and dogs. Missing items were diversity (five times), juridical advice (two times) and breed analysis (once). Veterinary literature (i.e. the term "prevention") was only sufficient to describe 17% (5/30) of the cats and dogs. Missing items where questions about heritability (13 times), education (10 times), diversity (5 times), juridical advise (2 times), breed analysis (once). In terms of the validity, all items in the definition from human medicine and "prevention" derived from veterinary literature where encountered in at least one of the cats and/or dogs.

For the second question, both the medicine- and veterinary literature-based description were sufficiently complete to describe all the cats and dogs. Nevertheless, in terms of validity, genetic counseling was never

Table 2 Comparing data from cats and dogs seen at the clinical genetics/genetic counseling service with the definitions found in literature

	Human medicine	Veterinary medicine	Missing items (human medicine)	Missing items (veterinary medicine)
What is genetic counseling?	79% (24/30)	17% (5/30)	Diversity (5x) Juridical advice (2x) Breed analysis (1x)	Heritability (13x) Education (10x) Diversity (5x) Juridical advice (2x) Breed analysis (1x)
Who provides genetic counseling?	100% (30/30)	100% (30/30)	No missing items	No missing items
Tools used in genetic counseling	100% (30/30)	0% (0/30)	No missing items	Information giving (30x) Psychological support (21x) Additional (not-DNA based) diagnostical tests (18x)

Overview of the extent to which the cats and dogs seen at the clinical genetics/genetic counseling service are completely described in the literature-based definitions from human medicine and veterinary medicine, respectively. The assessment of the completeness starts from the cats and dogs' point of view and evaluates whether the definitions adequately describe their situation. Any missing elements in the definitions are identified along with their respective frequencies

provided by a genetics researcher, as mentioned in the veterinary definition. While there was a close interaction between the veterinarian/veterinary genetic counselor and a genetics researcher in 4 out of the 30 feline/canine patients, the genetics researcher had no direct contact with the cats and dogs. Based on this data, this item is thus invalid.

For the third question, the medicine-based description was sufficiently complete to describe all the cats and dogs. The veterinary-literature based description never sufficed because there was no reference to information giving in veterinary literature. Even when this item, i.e. information giving, was not considered, only 23% of the cases were sufficiently described as there was a need to perform other additional (=not-DNA based) diagnostical tests in 18 out of 30 cases and additional psychological support in 21 out of 30. In terms of the validity, all items in the definition from medicine and from veterinary literature where encountered in at least one of the feline/canine patients, respectively.

Additional comparisons between veterinary literature and the dogs and cats.

Sixty-seven percent (10/15 articles) of the species mentioned in literature were dogs, 7% (1/15 articles) mentioned cats and in the remaining articles (4/15, 27%) the species was not specified. There was no additional signalment data, nor information regarding interdisciplinary collaboration. A detailed overview is provided in Supplementary File S1.

Based on signalment, 70% (21/30) of the patients were dogs, while 30% (9/30) were cats. They belonged to various breeds (cats: 5 Sphynx, 2 Persian, 1 Nebelung, 1 British Shorthair; dogs: 4 Border Collie, 3 Czechoslovakian Wolfdogs, 2 German Sheperds, 1 Scottish terrier, 1 Weimaraner, 1 Saint-bernard, 1 Labrador Retriever, 1 Husky, 1 Cane Corso, 1 Leonberg, 1 Chow Chow, 1 Old English Sheepdog, 1 Poodle, 1 Keeshond and 1 Samoyed dog). Both sexes were represented (11 males and 19 females, respectively). The youngest patient was a stillborn puppy and the oldest patient was a senior dog (aged 8 years old). Overall, the median age was 3 years. While 27% (8/30) patients purely visited the clinical genetics/genetic counseling clinic, 73% (22/30) were referred by or needed to be referred to at least one other discipline (i.e. medical imaging (12/30), cardiology (7/30), orthopedics (3/30), neurology (2/30), dermatology (1/30), ophthalmology (1/30), and/or nutrition (1/30), respectively).

Discussion

While genetic counseling in human medicine has become standard practice, literature on genetic counseling in veterinary medicine is rather scarce, eventhough genetic diseases are widespread in cats and dogs. While the ones that do mention it often share the expectation that genetic counseling will become a common practice, our literature search demonstrates that an actual description of what it entails, who provides it and what it is based on, is close to non-existent [20]. Furthermore, it was unclear whether what is mentioned in veterinary literature, accurately reflects the needs of cats and dogs. In other words, it was not clear whether that what is currently described in veterinary literature, is actually sufficient, complete and valid. It was neither clear whether the accepted definitions in medicine can simply be extrapolated to

the veterinary field. Correct definitions are however extremely important: amongst others, they ensure adequate treatment, correct diagnosis, optimal prevention, and reproducible research [14, 32, 33].

The aim here was to evaluate what has been published and eventually modify this to create definitions fulfilling the aforementioned criteria. To achieve this, a stepwise analysis was conducted. A first observation was that genetic counseling in human medicine is rather well-defined and accurately described by a limited number of reference works. A second observation was that in general, there are no true contradictions between veterinary literature and the definitions in human medicine. The veterinary literature however more or less consistently reports only a subset of what is mentioned in human medicine. This might reflect that only a subset is important in veterinary medicine. The only way to test this, was by comparing characteristics of cats and dogs with literature. This was possible thanks to the data from the clinical genetics/genetic counseling service and it seemed that feline/canine patient characteristics were overall more accurately described in the human definitions. A third observation was that what is cited in veterinary literature most often, is not necessarily what is needed or used most, which reflects discrepancies between the theory and the practice, as will be discussed in more detail when the specific questions are addressed.

A discussion on what genetic counseling exactly is, seems to be entirely focused on "prevention" in veterinary medicine. Veterinary literature is however scarce as it consists of only two publications (Table 1)(Fig. 1). An evaluation of the completeness revealed that a "prevention oriented" focus is indeed too narrow, as it only explained 17% of the cases, with five different items that were missing (Table 2). The much longer definition in medicine was however also not complete, as it only explained 80% of the cases (Table 2). That definition only missed three items, however. Starting from the most complete definition (i.e. the one from medicine), items on "genetic diversity", "juridical advice" and "breed analysis" might thus have to be added. From a validity point of view, every item mentioned in the definition on genetic counseling, was identified in the feline/canine group. In short, this indicates that the addition of three items has to be considered, while no items have to be removed.

The first item that might have to be added to the definition, is genetic diversity. It is known that genetic diversity is extremely important in veterinary medicine, as it is the cause of the high prevalence of genetic disorders, as well as the solution towards better health [34]. As such, we propose the addition of the word "genetic diversity" at two places (Table 3). The second item relates to questions regarding "juridical advice". While it came back on two separate occasions, subjectively, law-oriented questions do not truly seem to fit within the general scope of genetic counseling and might be a separate field. As such, to be on the safe side, we propose to not include it in the definition at this moment, keeping in mind that future refinements will likely be needed either way. The final item is "breed". While the situation is entirely different in medicine, breed-specific differences are mentioned in veterinary literature all the time and consistently taught

Table 3 Proposed definitions for genetic counseling in veterinary medicine

What is genetic counseling?	Genetic counseling is the process of helping animal owners and breeders understand – and adapt to – the medical, psychological, familial implications of genetic contributions to disease. This process integrates the following: - Interpretation of pedigrees and medical histories to assess the chance of disease occurrence or recurrence - Education about inheritance, testing, management, prevention, resources, research, and genetic diversity, taking breed-specific differences into account whenever applicable - Counseling to promote informed choices and adaptation to the risk or condition, while maintaining or improving genetic diversity
Who provides genetic counseling?	Veterinarians and veterinary genetic counselors with a specific training in the field
Tools used in genetic counseling	Tools: - essential for information gathering: e.g. family history (pedigrees) - used for establishing/verifying diagnosis: this includes DNA-testing, among other tests - related to risk assessment, i.e. they are used to calculate future reproductive or personal health risk - related to information giving, i.e. to help the animal owner and/or breeder understand the process and the implications of the findings for the affected animal and related animals - related to psychological counseling and support, e.g. to help the animal owner and/or breeder deal with emotional responses after giving distressing data

Proposed definitions for genetic counseling in veterinary medicine. The changes relative to the original definitions (in Table 1) are underlined

to take into account in veterinary training. As such, it seems no more than correct to add this term to the definition as well. Together with a few semantic changes (e.g. replacing the terms 'people' and 'family and medical histories' with 'animal owners and breeders' and 'pedigrees', respectively), a modified definition on genetic counseling is presented in Table 3.

The question on who provides genetic counseling, has been answered more often in veterinary literature than the first question on what genetic counseling is. While the vast majority of the answers are veterinarians, two articles leave it more open, with one specifying veterinary genetic counselors and the other genetic researchers. Overall, based on the feline/canine data, both veterinary literature and the definitions from medicine seem to be complete (explain both 100% of the cases). The item "genetics researcher" however was not found in our feline/canine data, as such, it seems to be an invalid item that might have to be removed, as discussed further.

Starting from the definition in human medicine, genetic counseling seems to be open to both medical doctors and specialists with other degrees (Table 1). The exact requirements seem to be more country-specific, however. In more detail, clinical geneticists are medical doctors that have completed an "Accreditation Council for Graduate Medical Education" (ACGME)-accredited residency in Clinical Genetics and Genomics and are boarded by the American Board of Medical Genetics and Genomics in the USA (or by boards in other countries) [35]. For genetic counseling, the situation is more complex. In the USA, genetic counselors hold a master's degree and and are boarded by the American Board of Genetic Counseling. In Europe, the Genetic Nurse and Counsellor Professional Branch of the European Board of Medical Genetics developed competencies and standards of practice for genetic counselor registration within the European Union [36]. The profession is established or under development in more than 11 European countries [36]. In 7 additional countries, genetic counselors are beginning to establish the discipline, but they were trained in other countries, however [36]. One example is Belgium. The profession of genetic counselor is not yet recognized there and there is no registration process yet. The genetic counselors practicing in Belgium have been trained outside of Belgium, some but not all at dedicated master in genetic counseling programs [37].

In veterinary medicine, only the veterinarian's degree is strictly defined. As such, this will be discussed first. There are some pros to having at least a veterinarian involved. In more detail, there were quite some feline/canine patients in which the diagnosis actually needed to be established (22/30 patients, also see Supplementary File S2). Both in veterinary medicine and in human medicine, diagnosing patients is, to our knowledge, by law restricted to veterinarians and medical doctors, respectively, so purely practically, a veterinarian seems indispensable. Studies have also shown that breeders and animal owners expect veterinarians to have in-depth knowledge about genetic testing and the interpretation of tests [38]. There is however a con: veterinarians do not always have the knowledge or have indicated themselves that they do not necessarily feel competent to interpret e.g. DNA-tests [39]. Parallel with the situation in human medicine, specific training thus seems a prerequisite to provide adequate services.

Whether genetic counseling should be restricted to veterinarians, is currently a hypothetical question as there are no alternative official programs at the moment. However, we do see the benefits to include a more diverse spectrum of professionals for the parts unrelated to diagnosis, as also mentioned in two veterinary articles and similar to the genetic counselor in human medicine [21, 22]. The genetic counselor has typically completed a relevant first degree (e.g. in nursing, midwifery, biology, psychology or social work), followed by a master's degree in genetic counseling which includes human genetics and counseling skills training [40]. As such, there are different professions that are for example more specifically trained in dealing with the emotional impact that a diagnosis might have, and this is also relevant in veterinary medicine as it was recognized in 21 out of 30 cases (Supplementary File S2). It remains thus to be seen how the profession evolves, but as the field continues to become more specialized, one can expect a similar evolution as in human medicine.

One specific item was only mentioned in veterinary literature, namely the involvement of genetics researchers, which is an interesting point of view [22]. They can certainly contribute thanks to their in-depth knowledge in the field of genetics. At the same time, being a research expert in genetics does not necessarily qualifies one to also adequately provide clinical services. This view also corresponds to our observation: while there was a close interaction between the veterinarian/veterinary genetic counselor and a genetics researcher in a subset of the cats and dogs, the genetics researcher had no direct contact with the animals. Combined, based on the absence of a genetics researcher from the definitions in medicine and as the item also was invalid based on our cats and dogs data, we propose to remove it at this moment.

Overall however, there seem to be quite some advantages to include professionals with a different expertise as together, they will provide the broadest overview from various angles and even now already, within veterinary medicine, the cases seen actually substantiate the advantage of a multidisciplinary approach as the majority of

the feline/canine patients involved a collaboration with other specializations.

Irrespective of who performs genetic counseling, it is of paramount importance that they are well trained in the tools involved in the field. As can be seen from the results in Table 1, the approach to describe them varies. While veterinary literature seems to work towards an exhaustive list, the reference works from medicine tend to group them under what their aim is and each time provide at least one example. Objectively, based on the evaluation of the completeness, it is clear that the latter method seems more adequate. Subjectively, this also seems to be a safer approach: it is difficult to provide a complete exhaustive list as techniques change and new ones are developed all the time. As such, we also suggest to adopt the approach from human medicine, with some semantic changes in line with what was also done for the definition on what genetic counseling entails (Table 3).

A discrepancy was found between what was cited most in veterinary literature and what was needed in the cats and dogs. In terms of frequency, DNA-tests only take up the third place in the latter (Supplementary File S2), while it is by far the most mentioned tool in literature (Table 1). While this was no surprise to us, it emphasizes the importance of a solid knowledge of other techniques beyond DNA testing by genetic counselors. The consultations placed for example significant emphasis on providing information to enable people to comprehend and manage conditions that are present in their animal's lineage. It also stresses the importance of having correct definitions. Just as the "prevention-oriented" view, a purely DNA test-oriented view also seems to tackle only a subset of what genetic counseling truly entails.

Aside from these main questions, we wanted to characterize the feline/canine patients for which genetic counseling is requested, based on their signalment. In medicine, the need for genetic counseling is various. In terms of the life stages involved, genetic counselors start to be involved prior to birth, during pregnancy, as parents tend to have non-invasive prenatal testing performed. This test allows geneticists to detect aneuploidy in the fetus [41]. Once a baby is born, s/he can suffer from birth defects or symptoms compatible with a genetic disease. Parents might need genetic counseling to understand what the different possibilities are in terms of treatment options and what their future will look like [42]. Genetic counseling is not only recommended in pediatric diseases, adults are being diagnosed with late-onset genetic diseases every day. Investigating what their own future will look like and/or what the risk for their progeny involves is primordial. Each stage of life has its own disorders and challenges. This is the reason why, in human medicine, genetic counseling is needed in a variety of specialisations [16]. Our observations indicate likewise a widespread age involvement, ranging from birth to seniority and similarly, the involvement of a wide range of other disciplines. In contrast with literature however, the demand seems to be present in both cats and dogs, while the veterinary literature is nearly entirely focused on dogs alone.

Generally, while this study seems to provide a good overview, there are some limitations that have to be discussed. In an attempt to reduce subjectivity while defining a new field, our methodology was based on criteria that should lead to a robust definition [14]. Moreover, all steps in the analysis were conducted independently and in parallel by two authors to minimize a potential bias of the evaluator. Nevertheless, the inclusion of more evaluators might have further reduced the chance of introducing a bias. There is also a limitation linked to the retrievability of literature: as some articles were inaccessible, even after attempts to contact the authors, some data might have been missing. Noteworthy, the unretrieved articles fall within the timeframe of 1993 to 2002, predating the task force of the NSGC's definition establishment for genetic counseling in human medicine in 2003. Given the subsequent exponential growth in tools and knowledge in the field, it is less probable that these inaccessible articles would significantly alter the study's outcomes. Another limitation is that the cats and dogs that were used to evaluate the definitions, were all seen at one clinic. This has the downside that they of course reflect the facility-specific situation and that one can question the generalizability of these results. Furthermore, we recognize the constraint of a limited sample size, comprising 30 dogs and cats, and acknowledge that this might also lead to a potential bias. While a larger sample size would undoubtedly be beneficial, various observations suggest that the field is actually reflected quite well overall and that it is thus unlikely that the aforementioned limitations have substantially influenced the results. Firstly, there is a close to perfect match with the original definitions that are used in human medicine and these are internationally agreed on, widely used and substantiated by a large caseload. Secondly, they do not contradict the limited veterinary literature, rather they encompass what is mentioned there. As such, we consider it rather unlikely that they are biased. Finally, we would like to stress that the current findings either way only serve as a foundation, laying the groundwork for further fine-tuning, and encourage others to participate in this process.

Overall, this study was conducted based on the observation that there is a considerable need for genetic counseling, while the field itself seemed to be ill-defined as no specific definitions were found, and a veterinary-specific evaluation of the definitions used in

medicine seemed to be lacking as well. We have found that genetic counseling in veterinary medicine closely resembles that in human medicine, with some specific adaptations, which confirms our prior hypothesis. Our goal is to help defining the field by providing some baseline definitions and encourage research efforts. As such, we also actively invite other parties in the field, to comment on these definitions, to collaborate to this branch in veterinary medicine and to join our veterinary genetic counselor network.

Conclusion

Dogs and cats have been subject to a great loss of genetic diversity over the years. This has resulted in a high prevalence of genetic disorders compared to humans. Where genetic counseling in human medicine is very important on an individual/familial level, it has stayed ill-defined in our domestic animals over the years. As we experience a high demand for genetic counseling in veterinary medicine and because this term is ill-defined, we propose here a definition for genetic counseling in small animal veterinary medicine, based on a literature search, an evaluation of cats and dogs presented at our clinical genetics/genetic counseling service and definitions in human medicine. Our definition is slightly different from the definition used in human medicine because animal breeding has unique characteristics that warrant specific attention, one of them being the more or less consistent issue of a lack of genetic diversity. Our aims by providing these definitions are to establish the groundwork for genetic counseling within veterinary medicine and inspire other researchers to develop this field into a distinct branch of veterinary medicine.

Supplementary Information

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Supplementary Material 1.

Supplementary Material 2.

Supplementary Material 3.

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Authors' contributions

Substantial contributions to research design, or the acquisition, analysis or interpretation of data: L.A, B.J.G.B, V.S, S.B, P.S, L.P, J.S. Drafting the paper: L.A., revising it critically: B.J.G.B. All authors read, improved and approved the final manuscript.

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Data availability

The datasets supporting the conclusions of this article are included within the article and its additional files. All data generated or analysed during this study are included in this published article (and its Supplementary Information files). Personal patient files are not provided to adhere to Regulation (EU) 2016/679 of the European Parliament and of the Council of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC (General Data Protection Regulation).

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Not applicable.

Competing interests

The authors declare no competing interests.

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