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Evaluating the Clinical Utility of Genomic Sequencing After Perinatal Death

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ABSTRACT

Following termination of pregnancy for fetal anomaly or unexplained perinatal death (PND), clinical geneticists advise on possible genetic causes and likelihood of recurrence, often with limited use of molecular analysis. In the Australian Genomic Autopsy Study (GAS) cases that were unresolved following standard-of-care investigations underwent exome and/or genome sequencing (ES/GS). This diagnostic before-and-after study measured the changes in clinical management, in terms of the effect on clinical counselling that was provided to parents following ES/GS. Clinicians were surveyed before and after receiving sequencing results about the likelihood of recurrence and the reproductive planning advice they would provide to families. 161 pairs of before-and-after surveys were completed. Clinician estimates regarding PND recurrence changed for 45% (73/161) of families after receiving test results, despite a genetic diagnosis being found in only 19%. Families with an '*unknown likelihood*' of recurrence reduced from 26% to 15% ($p = 0.01$). The information provided to parents about recurrence and reproductive planning increased significantly, both with and without a diagnosis, and clinicians reported that most parents expressed value was obtained from the investigation. The utility of genomic autopsy for clinical management is not restricted to families with a genetic finding.

1 | Introduction

Post-mortem investigations following a perinatal death (PND) or termination of pregnancy for fetal anomaly (TOPFA) routinely seek genetic abnormalities such as chromosomal and copy number variants. However, recommendations to investigate monogenic disorders are not consistent, and the availability of molecular sequencing of DNA following

Collaborators names and affiliations are listed in Appendix A at the end of the manuscript.

PND/TOPFA is often limited to phenotype-associated genes [1–5]. Untargeted exome or genome sequencing to identify causes of fetal anomaly or death, hereafter referred to as Genomic Autopsy (GA), is not yet standard care given the high levels of specialized expertise and additional costs that are involved.

The diagnostic capability of GA has been demonstrated; a recent systematic review identified 32 studies reporting over 2000 cases of perinatal death in which GA was provided, but while causative genetic variants were identified in nearly a third of cases, limited data on clinical follow-up was identified [6]. In the prenatal or neonatal critical care settings, diagnostic information will directly inform clinical care decisions for the fetus or child [7–9], but the clinical impact of genetic information after a pregnancy loss is less immediate. Following fetal anomaly and/or death, a clinical geneticist may discuss the potential for genetic or non-genetic causes with parents and may provide guidance on the likelihood of recurrence and relevant strategies for future reproductive planning. The importance of accurate information about whether the loss was associated with a genetic cause, the probability of recurrence, and the suitability of medical reproductive interventions for bereaved parents cannot be overstated. Prior studies have identified occasions where preimplantation or prenatal genetic testing became available following genetic diagnosis [6, 10, 11], but we are unaware of any study that systematically reports on the clinical interpretation and genetic counseling provided to bereaved parents, with and without GA. This study aimed to identify the impact that GA has on the clinical guidance provided to parents after a perinatal death, both for families that did and did not obtain a molecular diagnosis. This is an important consideration when identifying the role and value of this medical technology in clinical practice.

2 | Methods

Genomic sequencing in cases of PND and TOPFA, following a non-diagnostic chromosomal microarray was provided through the Australian Genomic Autopsy Study (GAS) commencing in 2016 through to early 2024 [10]. This publicly funded research study was approved by the Melbourne Health Human Research Ethics Committee as part of the Australian Genomics Health Alliance protocol (no. HREC/16/MH/251). The 'whole of study' series is yet to be published, however the initial findings (the first 200 cases) have been described previously, including detail on the study background and laboratory methods [10]. We report on a *sub-study* commenced in 2021 to assess whether GA changed the clinician interaction provided to bereaved parents. Genetic findings were reported according to ACMG Guidelines based on population frequencies, in silico pathogenicity predictions, sequence conservation scores, protein function and expression, and known disease associations [12]. Only variants classified as ACMG class 3–5 (variant of uncertain significance (VUS), likely pathogenic, or pathogenic) and relevant to the proband's phenotype were reported back to the referring clinician with detailed gene- and variant-level curation information to support interpretation of clinical utility.

3 | Survey Methods

To identify a *change* in clinical management in this diagnostic before-and-after study, comparative data was required. This was problematic as all families enrolled in the GAS received DNA sequencing. We therefore designed the sub-study to consist of 'before' and 'after' surveys paired at the individual case level, to identify the clinician assessment and counselling attributable to receipt of sequencing test results. Surveys asked clinicians to: (i) estimate the likelihood of PND recurrence for the parents (in the absence of reproductive medicine interventions) and (ii) identify relevant reproductive interventions, based on the available information at the time. 'Before' responses were based on family history, physical autopsy reports and any imaging/pathology available. 'After' responses were informed by the Mendeliome Report in addition to the previously available clinical information. Each survey also asked about the information discussed with parents, and/or the reason that information was not discussed.

Enrolment from within the overall series was unselected and sequential; the first survey was embedded into the initial Clinical Record Form completed by the referring clinician (generally a clinical geneticist) on GAS enrolment. After the transfer of DNA samples, analysis and generation of a Mendeliome analysis report, the same clinician was provided a secure electronic link and requested to complete a second survey ('after GA'). *Ad hoc* reminder emails were sent to clinicians who had not completed the 'after' survey to maximise survey response rates and representation.

To ensure data collection and consistency, surveys provided multiple choice answers and to reduce anchoring bias or the suggestion of Mendelian recurrence rates, the estimates of recurrence were presented in probability bands over 20% intervals (0 to <20%, 20% to <40%, 40% to <60%, 60% to <80% and 80% and above). Surveys also asked whether the proband's parents were provided with counselling around the likelihood of recurrence and reproductive medicine topics. When counselling was not provided, clinicians were asked why it was not provided—and in the before survey, specifically—clinicians could nominate by check-box that the reason counselling was withheld was 'waiting for GA findings'. The survey questions are available in Supporting Information: Item 1.

The 'after' survey also asked clinicians to recall of whether they had received any feedback from parents regarding value or concerns associated with the GA, and to broadly classify the nature of this. Questions and categories are presented in the final six survey questions. Due to the sensitive nature of the topic, the approved ethics protocol did not cover direct parent contact by the study team for data validation, therefore the findings concerning parent feedback are considered exploratory.

4 | Data Handling and Statistical Methods

The study data were collected and managed using REDCap electronic data capture tools hosted at the University of Adelaide [13, 14]. REDCap (Research Electronic Data Capture) is a

secure, web-based software platform designed to support data capture for research studies.

Data were analyzed using STATA Version 17 statistical software [15]. Pearson's chi-squared tests were used to compare survey responders and non-responders. Fisher exact tests were used to evaluate within-survey relationships, and McNemar tests were used to compare the changes in recurrence estimates and genetic counseling provided before and after reporting of genomic sequencing.

5 | Results

5.1 | Survey Participation and Population Characteristics

At the close of the study, matched pairs of surveys were available representing data for 161 affected fetuses or newborns (a diagram of the flow of study participation is presented in Supporting Information: Item 2). This represented a completion rate of 88% for the follow-up survey (184 cases had consented, successfully entered 'before' data, sequenced and received reports before study cut-off) and encompassed engagement with 38 different clinicians throughout Australia (more detail is provided in Supporting Information: Item 3). The average interval between the surveys was 7.1 months (range 1.6 to 17.3 months). The demographic characteristics of the study population are presented in Table 1, along with the summary GA results.

There were no major differences between baseline characteristics of affected individuals with complete and incomplete responses, except that a greater number without follow-up had prior genetic testing ($p < 0.03$), nor was there a significant difference in the distribution of genetic findings between the individuals with complete and incomplete response data. Non-responder data is shown in Supporting Information: Item 3.

A pathogenic or likely pathogenic (P/LP) variant that explained the cause of the anomaly/death was identified for 31/161 (19%) individuals, with 17 (55%) of these being de novo variants. Clinicians were informed of strong candidate variants of uncertain significance (VUS) warranting extended analysis in a further 15 (9%) fetuses, of which 13 were inherited and 2 de novo (reporting criteria described in Methods and Table 1 footnotes). The remainder either identified no relevant variants or candidate variants of greater uncertainty that did not reach the required threshold for diagnostic Mendeliome reporting and hence were not reported to the referring clinician at the time of the 'after' survey.

Before receiving GA findings, clinicians estimated that half of the families had a less than 20% likelihood of recurrence, indicating they were unlikely to be associated with an inherited genetic variant. They believed that a quarter of affected families had a Mendelian inheritance pattern. For the remaining quarter, there was too much uncertainty for the clinician to provide a quantitative estimate. This pattern differed only where there was prior family history or where affected individuals had previous molecular genetic testing ($p \leq 0.001$ for each); in these

TABLE 1 | Demographics of the fetal GAS cohort with before and after clinical management data.

Characteristic	Cases $N = 161$ (%)
Reason for referral	
Multiple congenital anomalies	93 (58%)
Single congenital anomaly	43 (27%)
Unexplained fetal or neonatal death	23 (14%)
Other	2 (1%)
Family history	
Isolated event for couple	129 (80%)
Recurrent event for couple	24 (15%)
Recurrent event for family	1 (1%)
Other	7 (4%)
Consanguineous parents	
Yes	4 (2%)
No	157 (98%)
Assisted reproduction	
Yes	21 (13%)
No	137 (85%)
Unknown	3 (2%)
Prior molecular genetic investigations	
None	135 (84%)
Gene, panel, or exome sequencing	24 (15%)
Unknown	2 (1%)
Sequencing[a] results	
Solved (P/LP variant)	31 (19%)
de novo variant	17 (11%)
Inherited variant	14 (9%)
Uncertain, strong candidate VUS and clinical team informed[b]	15 (9%)
de novo variant	2 (1%)
Inherited variant	13 (8%)
No variant reported	115 (71%)
Candidate variant; did not satisfy diagnostic Mendeliome reporting requirement[c]	8 (6%)
Unsolved; no candidate variant identified (extended genome sequencing pursued for some cases after this study)	107 (66%)

Abbreviations: GAS = genomic autopsy study; LP = likely pathogenic; P = pathogenic; PND = perinatal death; TOPFA = termination of pregnancy due to fetal anomaly; VUS = variant of uncertain significance.
[a]In this sub-series, all DNA samples were provided as family trios (88%), quads or quins, and the DNA regions sequenced were exome 57%, genome 37% and both (ES and GS) in 6% of cases.
[b]Candidate variants of uncertain significance (ACMG class 3) were only reported to the clinical team where the variant was predicted to be pathogenic by in silico pathogenicity prediction tools, absent or extremely rare in population databases and had sufficient clinical overlap to support a causation link of the gene/variant and the fetal condition.
[c]Candidate variants, that did not satisfy diagnostic reporting requirements, typically would not be reported to the clinical team unless further investigations generated additional evidence. Additional investigations occurred after the clinical management surveys were completed, that is clinical management surveys were completed as though no abnormalities were identified on initial ES/GS (grouped with the "unsolved" cases).

subgroups, clinicians generally expected higher recurrence rates (inheritance). The distribution of the initial estimates of recurrence (before GA) for all subgroups and by subsequent Mendeliome finding is presented in Supporting Information: Item 3. There was no significant difference in the distribution of estimated recurrence (before GA) between cases that were ultimately solved and not solved with GA ($p = 0.34$).

5.2 | Change in Estimates of Recurrence Following ES/GS

Across the whole cohort, the estimated likelihood of recurrence changed for 45% (73/161) of families (95% CI: 38%, 53%) upon receipt of the ES/GS findings. The average proportion of cases with a change in estimate of PND recurrence *per clinician* ($k = 38$) was 52% (95% CI: 39%, 65%). A summary of the paired before and after estimates showing the movement between categories of likely PND recurrence is shown in Figure 1, assuming likelihoods of recurrence ≥20% are associated with Mendelian inheritance.

After GA findings, the most common change (involving >20% of families) was that clinicians were initially unable, but became able, to estimate likely PND recurrence. The next most common change was the reverse, with clinicians becoming *unable* to estimate the liklihood of PND recurrence for 9% of families. There were also instances (9%) where the recurrence likelihood was revised from apparently inherited to not, and a small number (5%) of families, where clinicians increased the estimate of recurrence to reflect an inherited cause. Changes between estimates of recurrence pre- and post-GA occurred similarly for those with a P/LP variant identified (48%), a VUS identified (47%), or no relevant variants identified (44%).

The percentage of affected families with an '*unknown likelihood of recurrence*' was significantly reduced from 26% before GA, to 15% after GA results were provided ($p = 0.01$), while the percentage with an estimate of recurrence less than 20% (interpreted as *unlikely* to be affected by an inherited cause) increased from 50% before GA, to 65% after GA results were provided ($p < 0.002$).

The reduction in uncertainty was driven by affected individuals that had a genetic finding; for individuals with a positive finding, the proportion with an unknown recurrence likelihood reduced from 26% to 0% ($p = 0.008$), whereas for unsolved cases this decreased from 26% to 19% ($p = 0.15$).

The increase in estimates of *recurrence unlikely to be associated with inheritance* was driven by the sub-group with *no genetic finding*. Estimates of recurrence <20% increased from 51% to 68% ($p = 0.001$) for couples where no genetic cause was identified in their affected fetus/newborn, whereas for families where a genetic cause was identified, there was a smaller magnitude of change (from 48% to 55%, $p = 0.75$) associated with the identification of de novo P/LP variants.

Analysis of change in estimates by subgroups identified that change was more likely where there was a family history (changed estimates occurred in 66% of families with history vs. 40% of families without, $p < 0.02$). This was associated with the initially high proportion of clinicians unable to estimate a recurrence likelihood, then being able to do so after receipt of GA results. There was no indication that phenotype severity, affected organ system, assisted reproduction use, or previous gene analysis influenced the proportion of families with changed estimates of recurrence after GA (see Supporting Information: Item 3).

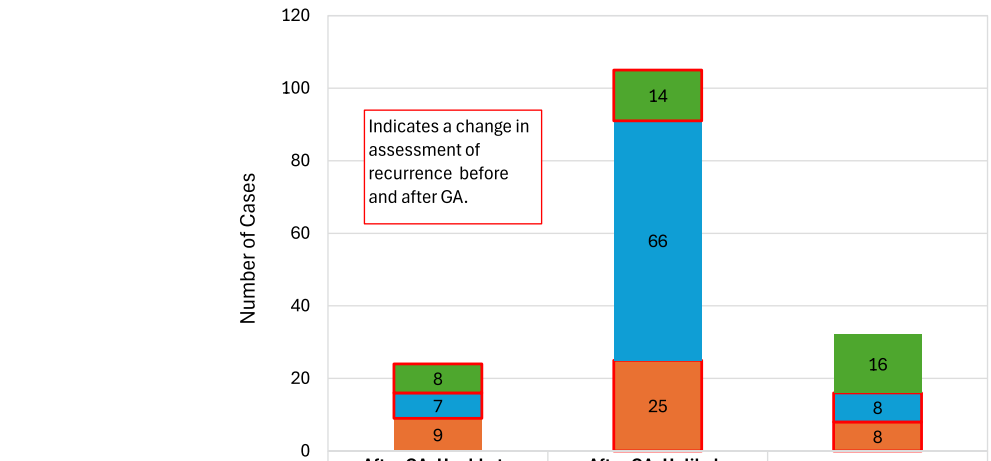

FIGURE 1 | Summary of clinician estimates of 'likelihood of recurrence' before and after genomic autopsy (paired dataset). Identification of a likelihood of recurrence greater than 20% is interpreted as indicating suspected Mendelian inheritance. The summary data differentiating all bands of recurrence likelihood as collected in the survey is shown in Supporting Information: Table SM3. [Colour figure can be viewed at wileyonlinelibrary.com]

5.3 | Change in Information on Recurrence Provided to Parents After ES/GS

The number of families where the available information was insufficient to guide reproductive counselling reduced significantly after GA (from 66% to 33%, $p < 0.001$), see Table 2. Before GA, clinicians in most cases (66%) considered the clinical information available to them was insufficient to guide reproductive counselling, and a further quarter were uncertain; however, clinicians considered the GA findings (alone or in combination with other investigations) were informative for most families (61%) and uninformative for fewer than one third of families.

There was a significant increase in the number of parents who were provided with counseling about the probability of recurrence: from 53% (85/161) of the parents before GA to nearly all, 94% (152/161) couples afterward, $p < 0.001$. Where an estimate was not initially provided: in 89% (68/76) of the cases, clinicians stated the reason was uncertainty; 4 clinicians said they were waiting for GA results, and other reasons applied in 4 cases. Counseling on the likelihood of recurrence was not always quantitative: 7 of the sets of parents before GA and 18 sets of parents after GA were not provided with a quantitative estimate.

5.4 | Provision of Specific Management Advice on Reproductive Planning

Both before and after genomic sequencing, clinicians identified reproductive planning (RP) topics as likely to be relevant to the parents of affected babies and identified whether they had provided counseling on those topics. A summary of the RP topics identified and those discussed with parents is presented in Figure 2.

RP information was considered relevant for 71% (115/161) of parents, but counselling or information was provided to only 50% (80/161) before GA (including 16 occurrences where it was not comprehensive). The reasons selected by clinicians as to why they did not discuss relevant RP topics were: that investigations had provided inadequate knowledge (69 cases); they were waiting for the outcome of GA (37 cases); it was not relevant to parents (2 families); and in another family, because the mother was already pregnant. After GA, clinicians were much more inclined to provide information on RP and discussed, or intended to discuss, RP topics with parents in 92% (148/161) of cases, although in some cases (6) the counselling after GA was incomplete. There was a highly significant reduction in clinicians citing they had 'inadequate knowledge' to provide counselling (from 43% before GA to 11% of the cohort after GA, $p < 0.001$), although this remained the most commonly cited explanation of why counselling was not provided or was limited (for 17 sets of parents). Other reasons were: parents were not interested (1), or unknown (1). Six of the 13 sets of parents where no counselling on RP topics was provided or intended to be provided after GA, had already received RP information prior to GA. Detail on the counselling topics by GA finding are presented in the Supporting Information: (Item 3).

5.5 | Exploratory Analysis of Parent Feedback to Clinicians

The parent feedback on GA, as recalled by clinicians, is presented in Table 3. There was no restriction on the number of different values or concerns that could be associated with parent feedback for each case. For most cases (100/161, 62%), clinicians recalled that parents had expressed that the GA investigation had been of value to them, predominantly by providing reassurance (45% of parents) or increased confidence (31%). However, clinicians also reported that a small number of parents (18/161, 11%) expressed concerns, most frequently reported as dissatisfaction that the investigation did not explain the loss and a sense of increased uncertainty (each reported by 6% of parents).

This preliminary data suggests a possible relationship between parents *expressing value* and the receipt of a genetic diagnosis: 84% (26/31) of parents of solved cases expressed value vs. 57% (74/130) of unsolved cases (1-sided Fisher's exact, $p = 0.004$). However, *no relationship* was observed between parents *expressing concern* and whether a genetic diagnosis was indicated ($p = 1.0$).

TABLE 2 | Responses to survey questions regarding adequacy of the information available to clinicians to inform counselling on reproductive planning (reporting paired before and after responses).

Before genomic autopsy		After genomic autopsy		
Does the available clinical information[a] provide sufficient information to guide reproductive counselling?		Are findings informative for counselling on future reproductive planning?		
		No	Uncertain	Yes
No	106 (66%)	39	6	61
Uncertain	39 (24%)	7	2	20
Yes	16 (10%)	7	1	8
Total	161	53 (33%) VUS 5, unsolved 48	9 (6%)VUS 2, unsolved 7	99 (61%) P/LP 31, VUS 8, unsolved 60

Abbreviations: P/LP = pathogenic/likely pathogenic; VUS = reported VUS.
[a]Clinical information in this context includes other investigations (microarray, tissue analysis, available family history, conventional autopsy, etc.).

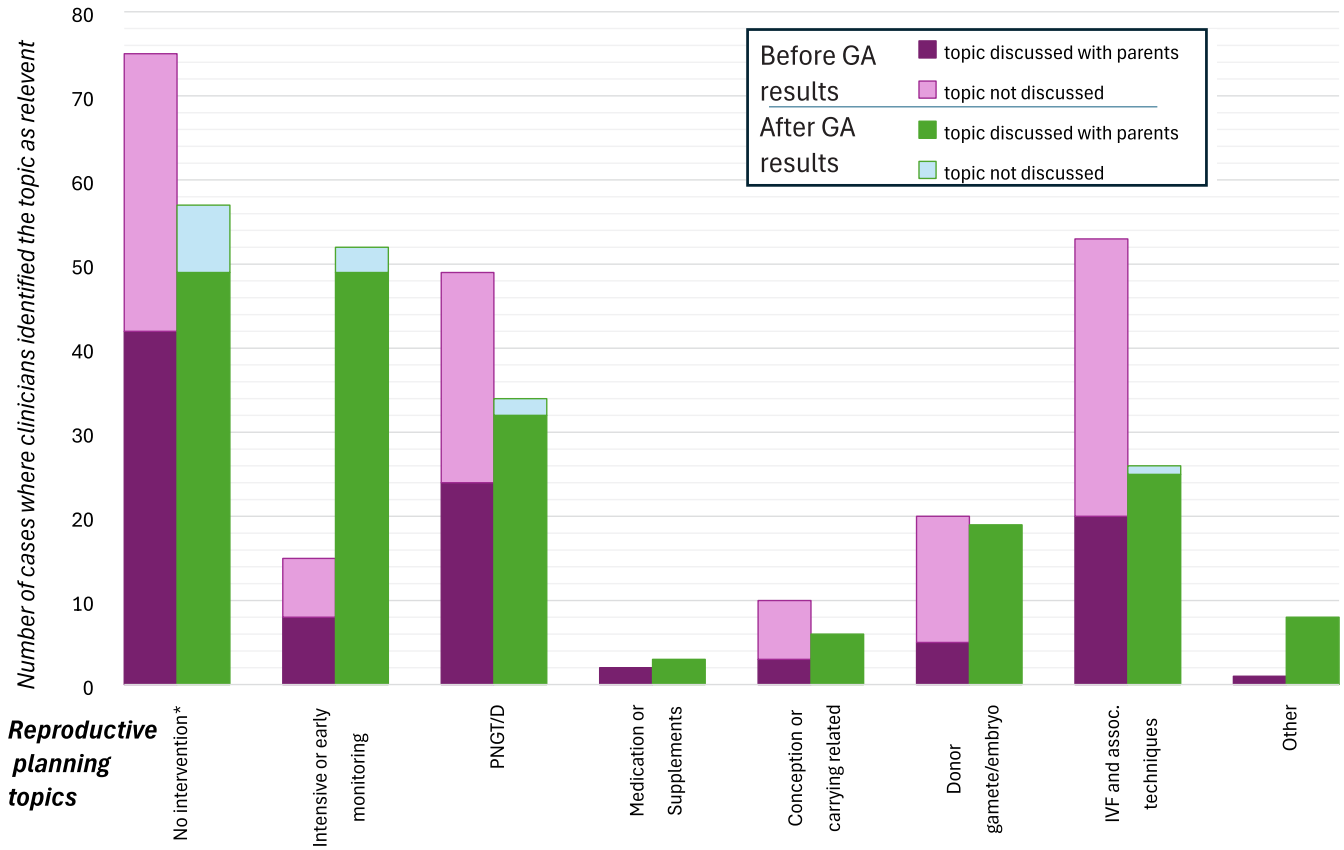

FIGURE 2 | Reproductive planning topics considered to be potentially relevant to parents, and whether counselling was provided (before and after GA). [Colour figure can be viewed at wileyonlinelibrary.com]

6 | Discussion

There are multiple interpretations of what constitutes 'utility' within the context of genomic testing [7, 16–18], and a single study is unlikely to comprehensively measure all aspects, even in a specific clinical application. In this study, we identified effects across three of the four domains of utility described by Hayeems et al. [17]; diagnostic thinking, therapeutic management and patient health/non-health outcome. Societal impact was not measurable by this study design.

For diagnostic thinking and the most immediate purpose of the investigation (determination of cause of anomaly/death), the rate at which GA can 'rule in' a genetic diagnosis provides an obvious measure of diagnostic utility. The diagnostic utility is less obvious for affected fetuses or newborns where no relevant variant is identified. The advice provided regarding interpretation of a 'no finding' result acknowledges that understanding of genetic variation is a field of ongoing scientific discovery and follows a Bayesian approach: no finding "reduces *but does not eliminate* the possibility that there is a genetic cause." [19] In this study, when interpreting estimated recurrence rates as a proxy for 'inherited' or 'non-inherited' diagnoses, diagnostic thinking was changed for nearly half of the families. The pre-test estimates of the likelihood of recurrence and the rate of diagnosis change were similar for cases that were subsequently solved and unsolved, indicating clinical markers and clinician judgement do not reliably predict inheritance status. When the proportion of cases with changed estimates of recurrence was calculated through weighting each clinician's data equally, the extent that GA findings changed the estimates of recurrence was slightly increased. This suggests the initial analysis approach may have yielded a conservative finding and confirms the significance of the findings is not due to a bias associated with the pattern of clinician involvement in the study. Of the cases with *no genetic finding*, the proportion designated as likely to be inherited decreased from 21% (24/115) pre-GA to 8% (10/115) post-GA. Sometimes, clinicians may effectively rule out a genetic diagnosis, but other times are less sure. In clinical practice, as expected, the interpretation of 'no finding' is variable and depends on factors other than a genomic result.

Progressing from a change in diagnostic thinking is the potential for change in management. In living patients, focus is given to accessing treatments, redirection of care, or prognosis [7, 16, 18, 20], but in the context of fetal autopsy investigation, clinical management relates to providing relevant RP information and strategies to prevent recurrence for parents. It is readily acknowledged that identification of a genetic cause (particularly when coupled with detail as to whether it is inherited or de novo) can facilitate the use of reproductive technologies (e.g., preimplantation or prenatal genetic testing) and ultimately prevent the recurrence of an affected pregnancy, and this is an important form of utility for families [7, 11, 16, 20]. In our study, all parents where an inherited variant was identified (9%) were provided with information about preventing recurrence with preimplantation or prenatal genetic testing or use of donor gametes. However, clinicians indicated that GA had been helpful for providing RP advice to many more parents (61%) than just those with a genetic diagnosis. This is consistent with a Delphi study across a broad range of clinicians

TABLE 3 | Clinician feedback on parent value or concern associated with the genomic autopsy.

		By genomic sequencing finding				
	All cases (*n* = 161)	**P/LP variant: inherited (*n* = 14)**	**P/LP variant: de novo (*n* = 17)**	**VUS: inherited (*n* = 13)**	**VUS: de novo (*n* = 2)**	**No variant reported (*n* = 115)**
Parents expressed that value was obtained						
Any value	100 (62%)	10 (71%)	16 (94%)	8 (62%)	1 (50%)	65 (57%)
Reassurance	72 (45%)	2 (14%)	14 (82%)	3 (23%)	0	53 (46%)
Increased confidence	50 (31%)	2 (14%)	15 (88%)	2 (15%)	1 (50%)	30 (26%)
Guidance	27 (17%)	6 (43%)	6 (35%)	4 (31%)	0	11 (10%)
Satisfactory explanation for loss	22 (14%)	7 (50%)	14 (82%)	1 (8%)	0	0
Enabled improved family planning	19 (12%)	8 (57%)	9 (53%)	1 (8%)	0	1 (1%)
Alleviated guilt	10 (6%)	2 (14%)	7 (41%)	0	0	1 (1%)
Other sentiments of value[a]	9 (6%)	1 (7%)	0	2 (15%)	0	6 (5%)
No value reported	61 (38%)	4 (29%)	1 (6%)	5 (38%)	1 (50%)	50 (43%)
Parents expressed that they had a concern						
Any concern	18 (11%)	2 (14%)	1 (6%)	3 (23%)	0	12 (10%)
Unsatisfactory explanation for loss	9 (6%)	0	0	1 (23%)	0	8 (7%)
Increased uncertainty	9 (6%)	1 (7%)	0	3 (23%)	0	5 (4%)
Increased anxiety	4 (3%)	2 (14%)	1 (6%)	0	0	1 (1%)
Limited family planning options	3 (2%)	0	0	0	0	3 (3%)
Increased sense of guilt	1 (1%)	0	1 (6%)	0	0	0
Other sentiments of concern[b]	3 (2%)	0	0	0	0	3 (3%)
No concern reported	143 (89%)	12 (86%)	16 (94%)	10 (77%)	2 (100%)	103 (90%)

Abbreviations: P/LP = pathogenic/likely pathogenic; VUS = reported VUS.
[a]Other expressions of value: "Pleased to have undertaken an investigation and opportunity involved in further study" (2 cases); "Grateful for information and options to further explore VUS even though uncertain at this stage," "Confidence progressing with donor embryo as no options for PNT or PGT."
[b]Other concerns: "Unable to have PGD for remaining embryos as they had so hoped," "Disappointment at no answers," "Did not want second pass of study, found the process of waiting too difficult."

who cared for infants/children, which concluded that ES impacts clinical decision-making for both cases with *and without* a genetic finding [21]. Although it appeared clinicians in our study mainly considered that the cause of anomaly/death was *unlikely* to be inherited (before the GA 50% predicted a recurrence rate < 20%), there was sufficient uncertainty among them that many did not feel confident discussing RP strategies, and the potential relevance of RP to manage an inherited condition was not able to

be dismissed. For example, more than 50% of clinicians considered information on IVF and associated procedures potentially relevant. Excluding natural conception and pregnancy monitoring, an average of 1.12 of the listed interventional RP topics were deemed likely to be relevant per couple, but after GA this decreased to an average of 0.78 topics per couple.

Presenting large volumes of information and multiple alternative options during genetic counselling can cause anxiety and confusion in parents [22], and our findings suggest that in the absence of GA, the volume of information with *potential but uncertain relevance* is an inhibitory factor to RP counselling discussions. The increased diagnostic confidence associated with positive or negative findings on GA enables clinicians to reduce the quantity of the RP information deemed relevant. It could potentially increase the quality of discussions. It was a limitation of this study that we were unable to directly capture parent feedback and relied on this to be communicated via clinicians, nevertheless our findings are reasonably consistent with a study of molecular sequencing in seriously ill children, which had a similar diagnostic yield (23%). In that study, 76% of parents reported they felt 'able to make informed reproductive planning or care for other family members' [23]. Clinician perceptions of utility were also highly concordant (81%) with parent's perceptions of test utility, with parents slightly more inclined to indicate utility [8, 23]. In our study, clinicians reported that most parents (62.1%, 100/161) had indicated that they found value in the GA investigation. We consider this is a likely underestimate for multiple reasons: (i) parent follow-up counselling was not complete with further counselling planned for at least 12 sets of parents; (ii) the study relied on parents spontaneously providing feedback to the clinician, which is highly dependent on personal inclination; and also (iii) we relied on the clinician's recall of parent feedback.

The survey provided clinicians an option of describing the value parents expressed from a list or entering free text. Some of the value parents identified appear directly associated with the availability of clinical management strategies (guidance and improved family planning), but around half (80/161) identified a value (reassurance, satisfactory explanation for loss or alleviated guilt) that is simply associated with 'having knowledge' rather than any clinical management or future outcome. Interestingly, one clinician noted, 'A negative result is rightly or wrongly often perceived as reassuring'.

A 'value of knowing' may be challenging to define but this study strongly suggests that 'knowing'—either of a diagnosis or that there is not an identifiable genetic diagnosis—is meaningful to a large proportion of parents following GA. Similarly, for a smaller proportion, there is possibly some *disutility* associated with the GA process. Concern was expressed by 10% (3/31) of parents with a genetic diagnosis (2 inherited, 1 de novo, all of which also reported positive personal value) and 10% (12/115) of parents where no findings were reported (of which four also identified positive personal value). The nature of the concerns differed among parents: parents whose babies received diagnoses expressed feelings of anxiety (from three sets of parents) or guilt (from one set of parents), which were directly linked to their knowledge of the condition. In contrast, parents of fetuses or newborns with undetermined causes of anomalies or death expressed disappointment over the lack of explanation. They experienced increased uncertainty and concern regarding the limited options in reproductive medicine. Their disappointment was related to unmet expectations or hopes, rather than the distress caused by diagnostic knowledge. These findings highlight the importance of managing expectations in pre-test counselling, reaffirmed by feedback from one clinician who noted parent response as 'Neutral, would have liked an explanation for loss, but had a low expectation of this'.

There are limitations associated with this study; we could not randomise allocation to GA and the fact that clinicians knew that they would get sequencing results may have influenced their initial (before) survey responses in unidentifiable ways. The prospective diagnostic before and after design (where clinicians are 'blind' for the before questions) minimised recall or confirmatory bias when identifying changes in the clinical guidance provided to parents. In addition, although enrolment in the sub-study was non-selective, there is potential for a self-selection bias for clinicians and/or parents participating in the broader study. The 38 different participating clinicians represent approximately 20% of the Australian national clinical geneticist workforce (estimated to be approximately 200 clinical geneticists in Australia in 2023) [24], which is a good representation, but does not exclude the possibility that findings could differ among non-participating clinicians. Clinical uncertainty may have been a factor in 'non-response', in which case our findings may be slightly reduced if follow-up was complete, however given the high participation rate, the impact of non-response would likely be small. A significant limitation is the reliance on clinician recall when reporting parent views leading to potential inaccuracies, and further research that directly elicits parent's values and concerns would be informative.

Reluctance to include GA as standard care following unexplained perinatal death may be associated with concerns regarding either the non-trivial healthcare resources associated with DNA sequencing, or the vulnerability of bereaved parents to further distress associated with investigations without findings. This study provides sound evidence that GA following unexplained perinatal death significantly enhances the genetic counselling provided to parents, both in cases where a genetic cause is identified and where no cause is identified; the diagnostic and clinical utility is greater than just the 'diagnostic yield'. Even where the cause of anomaly or death remains undiagnosed, clinicians generally have increased clarity and confidence in providing information around the likelihood of PND recurrence and future reproductive strategies. Preliminary feedback suggests these outcomes are broadly valued by parents and associated with a low rate of harm, irrespective of diagnosis. Clinicians and policymakers considering the role of this investigation can have increased confidence that, irrespective of the genetic finding, provision of GA as standard care after unexplained perinatal death can provide significant clinical value for bereaved families.

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Conflicts of Interest

The authors declare no conflicts of interest.

Data Availability Statement

The complete dataset associated with this study is not publicly available due to patient and clinician privacy restrictions. A partial dataset of relevant non-identifiable information may be available on request from the corresponding author.

Peer Review

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Supporting Information

Additional supporting information can be found online in the Supporting Information section.

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