

Newborn Screening Expansion and the Incidentally Detected Carrier: Parental Communication and Follow-Up Burden

Terhi Kangaslampi^{1*}, Rodrigo Villamediana-Ossa² and Elke Vanhauwaert³

¹Department of Newborn Screening, Vakka-Suomi Centre for Clinical Biochemistry, Uusikaupunki, Finland. ORCID: [0000-0001-1367-3536](https://orcid.org/0000-0001-1367-3536)

²Department of Clinical Genetics, Mancha Centre for Clinical Biochemistry, Albacete, Spain. ORCID: [0000-0003-9144-4151](https://orcid.org/0000-0003-9144-4151)

³Department of Genetic Counselling, Gaume Centre for Clinical Biochemistry, Virton, Belgium. ORCID: [0000-0001-1379-3896](https://orcid.org/0000-0001-1379-3896)

*Correspondence to: Terhi Kangaslampi, Department of Newborn Screening, Vakka-Suomi Centre for Clinical Biochemistry, Uusikaupunki, Finland, E-mail: t.kangaslampi@vscsb.fi

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Abstract

Expanded newborn screening panels using tandem mass spectrometry and increasingly genomic sequencing inevitably detect carrier status for some conditions alongside the affected infants the programme is designed to identify, and this incidental carrier detection generates a communication and follow-up burden that most screening programmes were not originally designed around. We reviewed carrier detection rates and downstream parental communication and follow-up activity across a national newborn screening programme's five-year experience expanding its panel, and surveyed parents who received an incidental carrier result about their understanding of its meaning and any reported anxiety. Carrier results were returned for 1.8% of screened infants across the expanded panel, and while 71% of surveyed parents correctly understood their child was not affected by the condition, 34% reported ongoing worry about their child's future health despite correct understanding, and 19% had sought additional, sometimes unnecessary, medical follow-up. We discuss what these findings imply for how carrier results should be communicated as panels continue to expand.

Keywords: Newborn screening; Carrier detection; Genetic counselling; Incidental findings; Parental communication

Introduction

Newborn screening occupies a distinctive position within preventive medicine: it is among the few population-wide screening programmes applied essentially universally to an entire birth cohort, generating an enormous volume of results, the overwhelming majority normal, with a comparatively small but operationally significant minority requiring further communication, follow-up, or action of some kind.^[1]

The success of newborn screening programmes in detecting treatable conditions early, before clinical presentation, has driven a sustained expansion of screened conditions over the decades since the earliest programmes began, with each additional condition added to a panel bringing its own incremental contribution to the overall volume of secondary, incidental, or otherwise unanticipated findings a programme

must manage, including, centrally for this review, carrier status findings that arise as an unavoidable by-product of screening for the affected state itself.^[2]

Newborn screening panels have expanded substantially over the past two decades, and tandem mass spectrometry-based screening for metabolic conditions, alongside emerging genomic sequencing pilots, detects carrier status for some conditions as an inevitable by-product of screening for affected infants, since the same biochemical or genetic signal that identifies an affected infant homozygous or compound heterozygous for a condition can also identify a heterozygous carrier who will never develop the condition themselves.^{[1][2]}

Carrier detection was not the original design intent of most screening programmes, and many were not built with a communication and counselling pathway specifically suited to

explaining a result that means the infant is healthy but carries a gene variant with potential relevance to future reproductive decisions or, in the case of some haemoglobinopathy screening, occasional mild clinical relevance.^[3] We reviewed one national programme's five-year experience with this pattern as its panel expanded, and surveyed parents about their understanding and any reported burden from receiving such a result.

This incidental carrier detection phenomenon sits within a broader category of incidental findings increasingly discussed across genomic medicine generally, but newborn screening presents a particularly distinct version of this challenge: the result is delivered to parents, not to the individual whose genetic status it concerns, at a point in that individual's life when they cannot participate in any decision about whether to receive the information, and often within the first days to weeks of the parents' own adjustment to a new child, a period during which parents' capacity to process complex genetic information may be limited regardless of how clearly that information is presented.^{[9][10]}

As genomic sequencing-based newborn screening pilots expand beyond the single-gene and targeted biochemical panels that have historically generated most carrier findings, the volume and diversity of incidental carrier results a programme must manage is likely to grow substantially, making the communication and resourcing questions this review examines increasingly pressing for programmes currently planning or piloting such expansion.

Programme Experience

Across the five-year period following panel expansion, carrier results were returned for 1.8% of all screened infants, the large majority relating to haemoglobinopathy and cystic fibrosis carrier status identified as a secondary finding of screening for the affected conditions themselves. Communication of a carrier result to parents was handled by a range of pathways across the programme's history, evolving from a standard letter in the programme's early expansion years to a structured phone consultation with a genetic counsellor for conditions considered to carry higher reproductive relevance.^[4]

Breaking down the 1.8% overall carrier detection rate by condition category, haemoglobinopathy carrier status accounted for 1.1 percentage points of the total, cystic fibrosis carrier status accounted for 0.5 percentage points, and the remaining 0.2 percentage points were distributed across several rarer metabolic conditions where carrier detection occurs less frequently given lower population carrier frequency for these specific conditions relative to haemoglobinopathy and cystic fibrosis.^[4]

The programme's communication pathway evolution over the five-year period reviewed was driven substantially by accumulating internal recognition, informed by informal parent feedback collected through the programme's family liaison service, that a standard letter alone was generating confusion and anxiety for a meaningful proportion of families, motivating the shift toward structured counsellor-led communication described

above for conditions the programme judged to carry higher reproductive or psychosocial relevance.

Parental Survey

A survey of 312 parents who had received an incidental carrier result in the preceding three years found that 71% correctly understood, when asked directly, that their child was not affected by the screened condition and would not develop it. However, 34% reported ongoing worry about their child's future health specifically related to the carrier result, including among parents who had correctly understood the child was unaffected -- indicating that correct factual understanding did not reliably prevent ongoing parental anxiety.^{[5][6]}

19% of surveyed parents reported having sought additional medical follow-up for their child specifically because of the carrier result, beyond what the screening programme itself recommended, including several reports of requesting repeat testing or specialist referral for a child who was, by definition, an unaffected carrier not requiring such follow-up.^[7] Parents who received a structured phone consultation with a genetic counsellor, rather than a standard letter alone, reported lower rates of both ongoing worry and unnecessary follow-up, though the programme's shift toward counsellor-led communication for higher-relevance conditions occurred over the same period as several other programme changes, limiting how confidently this specific association can be attributed to the communication method alone.^[8]

Breaking down the 19% unnecessary follow-up figure further, the most common specific action reported was requesting repeat testing for the carrier status itself, despite this status being stable and not subject to meaningful change, followed by seeking specialist paediatric consultation for symptoms the parent attributed to the carrier finding despite carrier status not being expected to produce clinical symptoms for the conditions represented in this programme's panel.^[7]

Parents of infants with haemoglobinopathy carrier results reported a somewhat higher rate of ongoing worry (39%) than parents of infants with cystic fibrosis carrier results (29%), a difference the survey could not definitively explain but that plausibly relates to differing parental familiarity with each condition, differing community-level awareness and experience with haemoglobinopathy in the specific population served by this programme, and differing framing in the communication materials used for each condition category over the study period.^[6]

Metric	Value
Carrier results (all screened infants)	1.8%
Correctly understood child unaffected	71%
Reported ongoing worry	34%
Sought unnecessary follow-up	19%

Table 1: Summary of Programme Carrier Detection Rate and Parental Survey Findings (n=312 surveyed parents).

Discussion

That over a third of parents who correctly understood their child was unaffected still reported ongoing worry suggests that the communication challenge here is not purely one of factual clarity -- many parents already grasp the distinction between carrier and affected status -- but of managing the emotional weight of a genetic result delivered in the first days of a child's life, often before parents have had time to process the broader experience of a new screening result of any kind.^[9]

As screening panels, and particularly genomic sequencing pilots, continue to expand the range of incidental carrier findings a programme will generate, the resourcing question this raises is not only whether results are communicated accurately but whether programmes have the genetic counselling capacity to manage the emotional and follow-up burden that accurate communication alone does not fully address; a programme scaling its screened-condition panel without proportionally scaling counselling capacity risks generating exactly the pattern of correctly-informed-but-still-anxious parents and unnecessary follow-up observed here.^[10]

Age and Family Context Considerations in This Survey

The 312 surveyed parents in this review's parental survey represented families whose child had received a carrier result up to three years prior, and briefly noting the family context characteristics of this surveyed group provides further useful background for interpreting the survey's findings.

Parents surveyed closer to the time of their child's original carrier result, within the first year following the result, reported somewhat higher rates of ongoing worry than parents surveyed toward the later end of the three-year recall window, suggesting that the ongoing worry this review documents may attenuate to some degree over time even without any specific additional intervention, though the survey design does not allow definitive conclusions about the trajectory of parental worry over time following a carrier result.^[6]

First-time parents were somewhat over-represented among those reporting both ongoing worry and unnecessary follow-up-seeking relative to parents who already had at least one older child at the time of the carrier result, consistent with the broader observation made elsewhere in this review that early parenting demands and relative inexperience navigating the healthcare system may compound the challenge of processing an unexpected genetic finding delivered in the newborn period.

Background on the Ethical Debate Around Incidental Carrier Detection

Incidental carrier detection through newborn screening has been the subject of a longstanding ethical debate within the newborn screening and clinical genetics literature, predating but directly relevant to the communication and resourcing questions this review examines, and briefly situating this review within that debate is useful context.

One strand of this debate concerns whether carrier status should be actively returned to parents at all, given that the child is not, by definition, affected by the condition and that the information's primary relevance is reproductive and therefore more directly pertinent to the child's own future reproductive decisions, decades away, than to the parents' immediate decisions about the newborn's care.^{[3][10]}

Most programmes, including the one examined in this review, have settled on returning carrier results to parents on the grounds that withholding a confirmed, accurate genetic finding raises its own ethical concerns and that parents generally want to know this information about their child even where its immediate clinical relevance is limited, a position this review's survey finding that most parents correctly understood their result once communicated is broadly consistent with, even though the same survey also documents the real psychosocial burden this disclosure can carry.

This review does not attempt to resolve the underlying ethical debate about whether carrier results should be returned at all, but rather takes as its starting point that most programmes have already settled on routine return of carrier results, and focuses specifically on how that return can be communicated more effectively given this settled practice.

Why Factual Understanding and Emotional Reassurance Are Distinct Communication Goals

This review's central finding, that a third of parents who correctly understood their child was unaffected still reported ongoing worry, points to a distinction worth making explicit for any programme designing or revising its carrier result communication approach: factual understanding and emotional reassurance are related but separable communication goals, and achieving the first does not automatically achieve the second.

A communication approach optimised purely for factual accuracy, clearly explaining carrier versus affected status, might reasonably be expected to maximise the 71% correct-understanding figure this review reports, but this review's finding that correct understanding and ongoing worry coexist in a substantial proportion of parents suggests that factual accuracy alone is an insufficient design target for a communication pathway intended to also address parental anxiety.^[9]

A communication approach additionally designed to address emotional reassurance would need to go beyond stating the factual distinction between carrier and affected status, to actively address the specific worries parents are likely to hold, concern about the child's future health despite correct understanding that the condition itself will not develop, concern about reproductive implications for the child's own future family planning many years hence, and general anxiety simply from having received any genetic result at all in the vulnerable early period after birth.

Regulatory and Guideline Context for Carrier Result Communication

Professional newborn screening and clinical genetics societies have issued guidance touching on carrier result communication, and briefly situating this review's recommendations relative to that existing guidance is useful for programmes seeking an authoritative complement to this review's own programme-based findings.

Existing guidance generally recommends that carrier results be communicated with factual accuracy and sensitivity to parental emotional response, broadly consistent with the direction this review's recommendations point, but existing guidance has generally been less specific about the concrete resourcing implications, how counselling capacity should scale with carrier detection volume, that this review emphasises as a central practical contribution.^{[2][10]}

We would encourage professional guidance addressing newborn screening expansion, particularly guidance specifically addressing genomic sequencing-based screening pilots, to incorporate explicit resourcing and workforce planning recommendations alongside existing communication-quality guidance, given this review's finding that communication quality and counselling capacity are closely linked in practice rather than being independent considerations a programme can address separately.

Timing of Communication Relative to Other Early Parenting Demands

The observation made in Discussion above, that carrier results are often communicated during a period when parents are still adjusting to the broader experience of a new child, deserves fuller exploration given its direct relevance to how and when communication should occur.

Carrier results from standard newborn screening typically become available within the first one to two weeks of life, a period during which parents, particularly of a first child, are simultaneously adjusting to sleep deprivation, establishing feeding routines, and processing the broader emotional and practical transition of early parenthood, a context not conducive to the kind of careful, attentive processing that a genetic result ideally warrants.^[9]

Some programmes have explored delaying carrier result communication by a short, defined interval beyond the point the result becomes available, reasoning that a brief delay, where clinically safe to do so, given carrier status carries no urgent clinical implication for the infant, might allow parents a somewhat less acutely demanding window in which to receive and process the information, though this approach has not been rigorously evaluated and this programme's own experience did not include a systematic trial of deliberate timing delay.^[4]

We would suggest that the timing question deserves dedicated future research attention, given that it represents a structural feature of communication, when rather than only how,

that this review's existing data cannot directly address but that plausibly interacts with the communication method questions this review does examine.

Specific Design Features of Effective Counselling Communication

Drawing on the better outcomes associated with structured genetic counsellor-led communication in this programme's experience, and on the broader genetic counselling literature, we outline specific design features that plausibly contribute to more effective carrier result communication beyond simply using a counsellor rather than a letter.

Active, two-way conversation allowing parents to ask questions in real time, rather than one-directional written communication, allows a counsellor to directly address the specific concern a given parent holds rather than providing a generic explanation that may not address that parent's particular source of worry, a feature a standard letter cannot replicate regardless of how carefully worded.^[8]

Explicit acknowledgement that ongoing worry despite correct factual understanding is a common and understandable response, rather than treating any residual anxiety as evidence of communication failure, may help normalise the parent's experience and reduce the sense that their continued worry reflects a misunderstanding on their part, a framing distinct from simply repeating the factual carrier-versus-affected distinction more emphatically.

Clear, explicit guidance on what follow-up, if any, is genuinely indicated for a carrier finding, and equally explicit guidance on what follow-up is not indicated, directly addresses the unnecessary-follow-up-seeking behaviour this review documents, by giving parents a concrete, actionable answer to the practical question of what they should do next, rather than leaving this question for parents to resolve through their own, sometimes unnecessary, independent follow-up.

Interdisciplinary Coordination Between Screening Programmes and Genetic Counselling Services

The communication improvements this review recommends depend substantially on effective coordination between the newborn screening programme itself, typically operating as a laboratory-based service, and genetic counselling services, which may be organisationally separate, and this coordination relationship deserves explicit comment.

Where genetic counselling services are organisationally separate from the screening programme, as was the case for part of this programme's five-year history reviewed here, establishing a reliable, well-resourced referral pathway between the two requires explicit institutional agreement regarding referral criteria, expected response time, and information sharing, rather than relying on informal, ad hoc coordination that may not reliably scale as carrier detection volume grows.^[8]

This programme's own shift toward structured counsellor-led communication for higher-relevance conditions required exactly this kind of explicit institutional coordination, and we would recommend that other programmes considering a similar shift begin by establishing this coordination agreement formally, with clear referral criteria mapped directly to the condition-level risk stratification discussed under Resourcing Implications above, rather than allowing the coordination relationship to develop informally over time as this programme's own did.

Role of Primary Care and Paediatric Providers in Reinforcing Communication

Beyond the newborn screening programme's own direct communication with parents, the child's ongoing primary care or paediatric provider represents a further point of contact where the carrier finding may be discussed, reinforced, or, in some cases, inadvertently miscommunicated, and this review's programme experience suggests this secondary communication channel deserves explicit attention.

Several of the unnecessary follow-up episodes reported in this review's parental survey arose from a primary care consultation initiated by the parent specifically to discuss the carrier finding, and in some of these consultations, according to parent-reported survey responses, the primary care provider's own explanation of the finding was reported by the parent as somewhat inconsistent with the explanation originally provided by the screening programme, potentially compounding rather than resolving the parent's uncertainty.^[7]

This pattern suggests that screening programmes should consider providing primary care and paediatric providers with their own dedicated reference material summarising the specific carrier finding, its clinical non-significance for the child, and the absence of any indicated follow-up, so that a parent raising the carrier finding in a subsequent primary care consultation receives a consistent, reinforcing explanation rather than a potentially inconsistent secondary account that could undermine confidence in the original screening programme communication.

Resourcing Implications for Programmes Scaling Their Panels

The resourcing argument made in Discussion above, that programmes expanding their screening panels should scale counselling capacity alongside the panel itself, deserves fuller elaboration given its direct practical implications for programme planning.

A screening programme's counselling capacity planning has historically been driven primarily by the volume of affected infants identified, a comparatively small and well-characterised number for most conditions, rather than by the volume of carrier results generated, which, as this review's 1.8% figure illustrates, represents a substantially larger absolute number of families requiring some form of communication and potential counselling support.^[2]

A programme planning to expand its panel, particularly toward genomic sequencing-based screening with its correspondingly larger potential carrier detection scope, should explicitly forecast expected carrier detection volume alongside expected affected-infant detection volume when planning counselling capacity, rather than scaling counselling resources based on affected-infant volume alone and treating carrier communication capacity as a secondary, residual consideration.

Where dedicated genetic counsellor capacity cannot be scaled to match growing carrier detection volume, a programme should consider a risk-stratified communication model, reserving structured counsellor-led communication for the conditions this review's own programme experience suggests carry higher reproductive or psychosocial relevance, while using a well-designed, carefully tested written communication format, incorporating the specific design features discussed above even within a written format, for lower-relevance conditions where full counsellor capacity cannot be justified.

Economic Considerations

The unnecessary follow-up documented in this review's parental survey carries a direct economic cost, both to the healthcare system absorbing the additional consultations and testing, and to families bearing any out-of-pocket cost or time burden associated with seeking this follow-up, and it is worth briefly quantifying this economic dimension alongside the psychosocial burden already emphasised throughout this review.

Nineteen per cent of 312 surveyed parents reporting unnecessary follow-up, extrapolated across the full population of carrier-result-receiving families over this programme's five-year experience, implies a substantial absolute number of avoidable consultations, repeat tests, or specialist referrals accumulated across the programme's full catchment population, representing a real, if likely modest per-case, economic cost that a well-designed communication approach could plausibly reduce.^[7]

We would argue that the economic case for investing in improved communication, specifically the structured counselling approach and reinforcing primary care materials recommended throughout this review, rests not only on the psychosocial benefit to families but on this avoidable downstream healthcare utilisation cost, strengthening the case for programme investment in communication quality beyond a purely compassionate or patient-experience rationale alone.

Comparison with Previously Published Newborn Screening Communication Studies

This review's findings are broadly consistent with, and extend, a prior literature examining parental psychosocial response to newborn screening results more generally, including both false-positive results and confirmed carrier or affected findings.

Prior studies examining parental anxiety following a false-positive newborn screening result, a related but distinct

scenario from the confirmed carrier finding examined in this review, have similarly found that anxiety can persist even after a false-positive result is subsequently clarified as not indicating disease, suggesting a general pattern across several types of unexpected newborn screening result where correct eventual understanding does not fully resolve the emotional impact of having received an initially concerning result.^{[5][6]}

This review's contribution relative to that prior literature is to document this same general pattern specifically for the confirmed, stable carrier finding scenario, where, unlike a false-positive result, there is no subsequent correction to deliver, since the carrier finding itself is accurate and stable, meaning the ongoing worry this review documents cannot be addressed through the kind of result-correction communication that can at least partly address false-positive-related anxiety.

Why This Review Combines Programme Experience With Survey Data

This review deliberately combines two distinct evidentiary sources, the screening programme's own five-year operational experience and a dedicated retrospective parental survey, and it is worth briefly explaining why this combined approach was judged more informative than either source alone.

The programme's operational experience alone could establish the carrier detection rate and document the programme's own evolving communication practice, but could not, without the dedicated survey, directly measure parental understanding or psychosocial outcome, the two measures most directly relevant to assessing whether a given communication approach is actually achieving its intended purpose.^[4]

The parental survey alone, without the programme's operational context, could document parental understanding and reported worry but could not situate these findings against the specific communication pathway each surveyed parent actually received, the comparison between standard-letter and counsellor-led communication that forms this review's most actionable finding regarding which approach is associated with better outcomes.

Combining both sources, while introducing the confounding limitation already acknowledged under Study Limitations, allowed this review to connect programme-level communication practice directly to family-level outcome in a way neither source alone could achieve, and we would recommend this combined operational-and-survey approach to other programmes seeking to evaluate their own carrier result communication practice.

Equity Considerations in Communication Access

The structured counsellor-led communication approach this review's programme experience associates with better outcomes is, by its nature, a more resource-intensive communication channel than a standard letter, and it is worth addressing directly the equity implications of a risk-stratified communication model that reserves this more resource-intensive channel for only a subset of carrier findings.

Any risk-stratification approach necessarily means that families receiving a carrier result for a condition judged lower-relevance will receive a less resource-intensive communication experience than families receiving a result for a higher-relevance condition, and programmes adopting this approach should ensure that the lower-relevance communication pathway still meets a defined quality standard, incorporating the effective-communication design features discussed above even within a written format, rather than treating lower-relevance-condition families as a lower priority for communication quality generally.^[8]

Programmes should also consider whether their risk-stratification criteria for which conditions warrant counsellor-led communication versus a written approach adequately reflect variation in parental familiarity and community-level experience with specific conditions, given this review's own finding of differing worry rates between haemoglobinopathy and cystic fibrosis carrier results, rather than applying a single, fixed risk-stratification criterion uniformly across all communities served by the programme.

Study Limitations

This review's findings should be interpreted alongside several limitations.

The parental survey was conducted retrospectively, up to three years after some parents received their carrier result, introducing potential recall bias regarding both their understanding at the time and their subsequent follow-up-seeking behaviour, and the reported associations between communication method and outcome should be interpreted as describing a pattern observed within this programme's specific experience rather than as a controlled comparison isolating communication method from the other programme changes occurring over the same five-year period.^[4]

This review draws on a single national programme's experience, and both the specific carrier detection rate and the specific survey findings may differ at other programmes with different population carrier frequencies, different panel composition, or different existing counselling infrastructure and communication conventions.

Applicability to Other Incidental Findings Beyond Carrier Status

Although this review has focused specifically on incidental carrier detection, newborn screening programmes, particularly those piloting genomic sequencing approaches, increasingly generate other categories of incidental finding beyond carrier status, and briefly noting how this review's conclusions might extend to those other categories is useful context.

Incidental findings of adult-onset condition risk, variants of uncertain significance, and findings unrelated to the condition originally being screened for each raise communication challenges that share some features with the carrier detection scenario examined in this review, particularly the challenge of communicating a result with no immediate clinical relevance to the newborn but potential long-term or family relevance, but each also raises its own distinct ethical and communication considerations not fully captured by this review's carrier-specific findings.^[3]

We would expect the general principle this review emphasises, that factual accuracy and emotional reassurance are distinct communication goals requiring deliberate, separate design attention, to extend to these other incidental finding categories, though the specific communication content and resourcing requirements would need dedicated study for each category rather than assuming direct transferability of this review's carrier-specific findings.

Future Directions

A prospective, randomised or quasi-randomised comparison of structured counsellor-led communication against a well-designed written communication format incorporating the specific design features discussed above would provide stronger evidence than this review's retrospective, confounded comparison can offer for which specific elements of communication approach drive the better outcomes associated with counsellor-led communication in this programme's experience.

As genomic sequencing-based newborn screening pilots continue to expand, prospective monitoring of carrier detection rate and associated parental psychosocial outcomes, following this review's own methodology, at programmes adopting this broader screening approach would help establish whether the communication and resourcing challenges this review documents for biochemical-panel-era carrier detection scale proportionally, or disproportionately, as the range and volume of potential incidental carrier findings grows with genomic screening adoption.

Practical Checklist for Programmes Reviewing Their Communication Approach

As a closing practical summary, we offer the following checklist for a newborn screening programme director reviewing or designing a carrier result communication pathway.

Forecast expected carrier detection volume across the full panel, not only affected-infant volume, when planning genetic counselling capacity, particularly before expanding toward genomic sequencing-based screening with its larger potential carrier detection scope.

Design communication materials and pathways to address both factual understanding and emotional reassurance as distinct goals, incorporating explicit acknowledgement that ongoing worry despite correct understanding is common, and explicit, concrete guidance on what follow-up is and is not indicated.

Provide primary care and paediatric providers with dedicated reference material summarising each carrier finding type, to support consistent reinforcing communication when parents raise the finding in a subsequent consultation.

Where full genetic counsellor capacity cannot be scaled to match growing carrier detection volume, adopt a risk-stratified communication model, reserving counsellor-led communication for higher-relevance conditions while applying the specific effective-communication design features discussed in this review even within a written format for lower-relevance conditions.

Conclusion

Carrier results were returned for 1.8% of infants across an expanded newborn screening panel over five years, and while most surveyed parents correctly understood their child was unaffected, a third reported ongoing worry and nearly a fifth sought unnecessary follow-up. Structured genetic counselling communication was associated with better outcomes than a standard letter alone. Screening programmes expanding their panels should scale counselling capacity alongside the panel itself, since accurate factual communication does not on its own prevent the anxiety and follow-up burden an incidental carrier result can generate.

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