Title: A qualitative study exploring family communication following a diagnosis of adrenoleukodystrophy in the UK.

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Background:
X-linked adrenoleukodystrophy (ALD) is a progressive neurodegenerative condition with a variable clinical spectrum. Without early intervention, childhood cerebral ALD can result in rapid deterioration and death within a few years. Effective family communication is essential for identifying other relatives who might be at risk following a diagnosis. While there is a significant body of literature exploring family communication of genetic information, these studies cover a limited number of conditions, including hereditary cancer predisposition syndromes and inherited cardiac conditions. ALD has a unique set of disease characteristics that are likely to impact upon family communication but as of yet remain largely unexplored. This qualitative study aimed to explore the psychosocial impacts of receiving an ALD diagnosis and consider their implications for family communication.

Research Question
What are the experiences of those who receive a genetic diagnosis of adrenoleukodystrophy and how does this impact upon family communication?

Aims
The aims of the study were to:

- Explore the psychosocial impacts of receiving an ALD diagnosis.
- Explore the experiences of those who have received an ALD diagnosis for either themselves or their children in communicating genetic information to their relatives.
- Explore perceptions of how genetic counsellors might best facilitate the process of family communication following an ALD diagnosis.
- Establish an evidence base that can be used to help guide future genetic counselling practice.

Methods
Participants were recruited through the UK patient support group Alex, The Leukodystrophy Charity (Alex TLC). Seven semi-structured interviews were conducted with mothers of diagnosed children (n=4) and adult males who had received their own diagnosis (n=3). Interview data was analysed using reflexive thematic analysis.

Results
Three overarching themes were identified in the data: (1) The vicissitudes of diagnosis (2) Negotiating new roles and responsibilities and (3) Changing relationships.

> **The significant impacts of diagnosis:** All participants recounted the diagnosis of ALD to be a life changing experience. For many it was the point at which their world turned upside down and their goals and priorities changed. The diagnosis was described as a period of compounding stressors which took participants to their lowest point. Taken together, feelings of shock, denial, guilt, fear, uncertainty and loneliness produced a scenario of almost overwhelming grief.
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> **Negotiating new roles and responsibilities**: Despite this, participants described having to negotiate the new roles and responsibilities that had been bestowed upon them. As the first recipient of new genetic information within their family, they often took the lead on informing their relatives of the diagnosis and protecting them from its potential impacts. However, most found this to be both a difficult and daunting task, and a range of factors influenced how they approached the situation.

Changing relationships: Following a diagnosis, relationships with friends and family often changed. For some, the period of adversity fostered growth and connection, while for others, the weakening of existing relationships added to the overall stress of the situation.

Conclusions and the implications for genetic counselling practice

This is the first study to demonstrate how the severe mental and emotional toll of an ALD diagnosis can directly impact upon an individual's ability to effectively communicate genetic information to their relatives. Considering the potentially severe implications for non-disclosure to at-risk relatives, clinicians have an important role to play in facilitating the family communication process.

Results suggest that clinician support is likely to be most effective when offered in stages, with a shift in focus as patients move forward on their ALD journey. Crisis counselling with a focused short-term approach may support individuals to overcome the intense shock of the diagnosis and reconcile with their trauma. Following this, gradual and regular post-consultation contact would then enable a greater focus on family communication and reduce the burden that patients and/or their parents feel to immediately assume the role as expert in the condition at such a challenging time. In addition, particular attention should be paid to addressing feelings of motherly guilt which can directly hinder effective family communication. Healthcare professionals should consider the approach and timing of informing mothers of their carrier status as this has been shown to engender and exacerbate feelings of blame.

This study demonstrated that the communication challenges that ALD families face are not uniform. They are influenced by a range of factors and can occur at different stages post diagnosis. For example, while one participant recounted difficulties in accurately recalling genetic risk information in conversations with her relatives, another described how she and her husband were unable to reach an agreement on how to best proceed in the face of adversity. While these challenges are both centred in communication, effective solutions are likely to take different forms. The former suggests that bespoke communication aids would help some individuals to accurately disseminate risk information provided to them. The latter suggests that a mediation role which facilitates families to discuss challenges as they arise could be the more effective strategy in other families. Ultimately, results suggest that clinicians should not offer a 'one size fits all' approach to supporting family communication but instead collaborate with the ALD families they see to develop communication support strategies which are relevant to the challenges that they are likely to face.

Throughout the diagnostic period, healthcare professionals should ensure that they are providing accurate and detailed information about the condition. Patient support groups such as Alex TLC are well placed to support in this. Although information about cerebral onset is always going to be challenging, ensuring that patients do not stumble across these details without prior supportive discussion is likely to reduce their trauma.

For some, it appears that being tasked as the information bearer is too great a challenge given the devastating circumstances they are facing. This suggests that are more family-focused approach, in which the burden does not rest on a single individual or couple to hold responsibility to their relatives, might be helpful. Approaches such as genetic counsellor-led multi-family discussion groups might foster a supportive atmosphere and facilitate emotional disclosure between relatives in ALD families.

This study has made tentative suggestions about the types of interventions that would likely benefit ALD families in supporting family communication. Further research is now required to garner opinion on the types of intervention that are considered the most useful. This could lay the foundations for trialling a particular intervention with a sample group to determine its effectiveness.