The challenges for patients and sonographers when complex obstetric anomalies are identified.

Abstract

The prenatal identification of complex obstetric anomalies can present issues for expectant parents, sonographers and departments of obstetric ultrasound. The limitations of imaging technologies, ongoing fetal development and various interpretations of anomalies and prognosis create ambiguity. Complexity is further heightened by a lack of policies and training related to the communication of unexpected findings and support for sonographers who work with expectant parents during or after times of significant distress and trauma. This case report details challenges presented upon a third-trimester diagnosis of a complex obstetric anomaly; agenesis of the corpus callosum and offers recommendations to reduce the negative psychosocial consequences of complex obstetric anomaly diagnosis.

Introduction

Obstetric ultrasound provides an opportunity to assess the health and development of a fetus to allow parents to make informed reproductive decisions. [1] The prenatal identification of complex obstetric anomalies can present challenges for expectant parents, sonographers and ultrasound departments. Agenesis of the corpus callosum (ACC) is one such anomaly which is particularly complex due to variations of terminology, sonographic appearances and prognosis. While ACC is a neurological anomaly classified by the absence or underdevelopment of the corpus callosum (CC), the definitions of the terms such as complete or partial agenesis, dysgenesis, dysplasia, hypoplasia and hypogenesis overlap or conflict between reports, which increases the ambiguity within the diagnosis. [2, 3]

ACC is challenging to diagnose prenatally using ultrasound. Direct non-visualisation of the CC is only possible on a midsagittal view of the fetal brain, which is not a screening view typically included in morphology scan protocols as per international guidelines. [4] A novel technique through an axial plane was proposed but is not standard. [5] ACC may be suspected through indirect signs as outlined in Table 1 however, atypicality may be hard to identify before 24 weeks.[6] Furthermore, the lateral ventricles at mid-gestation may measure under 10mm, despite ACC. Within a study of 135 fetuses with CC anomalies, dilation occurred in 39/67 (58%) assessed before 24 weeks gestation. [9] Progressive ventricular dilation during gestation was common, with ventriculomegaly reported in 85% of cases at late gestation and hence considered part of the ACC presentation. [9] Progressive dilation likely accounts for the frequency of ACC diagnoses made after the mid-gestation scan. A report of a decade of cases included 26% identified incidentally during a third-trimester scan. [10] Given these difficulties, it is unlikely that a sonographer would be able to confirm a diagnosis of ACC at the time of a routine morphology scan but may identify an atypical appearance. The suspicion of ACC during a morphology scan, warrants tertiary referral for further investigation. [11] Further investigations may delineate whether the ACC is isolated or complex which may provide specific prognostic information. [2, 11]

The neurodevelopmental outcomes after a prenatal diagnosis of ACC vary from typical development through to disability with high support needs, and less frequent fetal or neonatal demise. [2] With a broad spectrum of outcomes and further investigations warranted, the

communication of a suspected ACC finding to expectant parents is likely to be difficult and stressful for the sonographer. Similarly, receiving unexpected news can be traumatic for expectant parents, and their experience of prenatal diagnosis and pregnancy may be influenced by the initial communication. This case report details the challenges that presented with a third-trimester diagnosis of a complex obstetric anomaly; agenesis of the corpus callosum and offers recommendations to reduce the negative psychosocial consequences of complex obstetric anomaly diagnosis.

Case Description

A 30-year-old primigravid woman was referred for a third trimester at 31 weeks and 0 days gestation due to pelvic pain. The woman had an unremarkable medical history, had declined first-trimester screening and had undergone a routine 20 week morphology scan which was reported as normal. The scan at 31weeks and 0 days showed satisfactory growth with no cause of pelvic pain identified. An incidental finding of dilated lateral ventricles measuring 19mm was noted (Figure 1). The remainder of the intracranial and wider fetal anatomy revealed no abnormality. The woman learned of this finding by reading her report as she was not informed by the sonographer or her referring doctor. With no one to discuss the findings, she searched the internet to gain understanding.

After a self-referral to Maternal Fetal Medicine (MFM), the woman underwent a transabdominal and transvaginal scan at 32 weeks and 5 days revealing bilateral ventriculomegaly of 18–20mm and non-visualisation of the CSP. At the conclusion of the scan, the sonologist confirmed the ventricular dilation and stated that the CC could not be visualised, but an absence could not be confirmed due to the limitations of the scan. The option of termination of pregnancy (TOP) was presented, with the suggestion that the woman could tell people she experienced a stillbirth. This was unexpected and presented before undergoing further investigations or meeting with the MFM team later that day. The woman was offered further investigations at her request and was informed that testing would not be cost-effective if she chose a TOP. The woman elected to undergo amniocentesis and was counselled about the prognosis of severe ventriculomegaly from a geneticist, obstetrician and paediatrician.

Fetal MRI performed at 33 weeks and 6 days (Figure 2) identified severe commissural dysgenesis with complete ACC and only a tiny residual anterior commissure present. Prenatal counselling at 35 weeks was provided with the revised diagnosis of isolated ACC with secondary colpocephaly, with an improved prognosis from that presented earlier. After the appointment, on the drive back to her regional home, MFM specialist informed the woman by phone that there were varied impressions among the MFM team including a higher likelihood of poor outcomes due to the level of commissural dysgenesis. Aicardi syndrome was presented as a significant possibility.

Upon choosing to continue her pregnancy, the woman was referred to her local regional hospital for the remainder of her antenatal care and birth. During an ultrasound at 37 weeks and 2 days (Figure 3), she explained her baby's diagnosis to the sonographer. While scanning, the sonographer stated the fetus had hydrocephalus and asked the women why she did not terminate, adding that in their country of birth, people would do so. This question increased the distress of the woman, who was alone in the appointment. The weight of the earlier decision making had brought sleep disturbance, nightmares, fear and sadness. The emotions and somatic symptoms, which had begun to ease after the decision to continue the pregnancy, returned with the sonographer's question.

The woman delivered a health female infant at 39 weeks and 6 days following an induction of labour. The diagnosis of ACC was established by cranial ultrasound at one-day-old and confirmed by MRI at three months of age (Figure 4). The ultrasound at one-day-old was undertaken by the same sonographer the woman saw weeks earlier and triggered difficult emotions. The woman experienced poor mental health requiring intervention in the post-natal period.

Discussion

This case of a third-trimester diagnosis of ACC for a regional mother presents several challenges that present for expectant parents, sonographers and departments of obstetric ultrasound.

Challenges for expectant parents

Many parents attend prenatal ultrasounds with the hope to determine the sex of the baby or for the opportunity to bond, and as such are ill-prepared for the news of an unexpected finding which can be shocking, and often traumatic. [12, 13] Post-traumatic stress disorder (PTSD) is highly prevalent among women who have experienced a prenatal diagnosis. [14, 15] Two common symptoms of PTSD after a prenatal diagnosis include the sense of re-experiencing the trauma or the fear and helplessness and hyperarousal. As such, sonographers interacting with patients who have experienced unexpected findings in their current or previous pregnancy should be mindful that undergoing another ultrasound or revisiting the same clinic may trigger symptoms of PTSD and distress, as exemplified in the case. This is of heightened relevance within regional or remote areas where patients may not have the option to attend a different clinic, and where departments may not be able to offer alternative obstetric sonographers if requested.

The imprecise nature of prenatal diagnosis creates uncertainty and the potential for diagnostic and prognostic change over the gestation. Human factors, such as the use of ambiguous terms or varying interpretations can mean that parents are told different information by different providers, as demonstrated. Furthermore, regional and remote parents often have to travel to MFM clinics, where providers are less likely to have links with the parent's local organisations, complicating referral pathways for ongoing support. While ongoing MFM follow up and ultrasounds after a diagnosis ACC may be within the management protocol [10], this case study highlights that this may not occur for regional patients.

The communication of a prenatal diagnosis and within follow up appointments can influence parent experience. [16-18] This means that sonographers have the potential to positively impact parental outcomes related to mental health and wellbeing. Common psychological responses and recommendations to reduce distress are outlined in Table 2.

Challenges for sonographers

Within Australia, the responsibility of the communication of unexpected findings in obstetric ultrasound has increasingly, yet inconsistently, shifted to sonographers. [19] A survey of qualified obstetric sonographers and trainees found that 79 (31.7%) respondents had communicated a congenital anomaly finding to their pregnant patient within the preceding month. [20] Despite this frequency, 63.5% of workplaces did not have policies about the communication of unexpected findings, and only 35% of the respondents had undertaken specific training, most commonly after qualifying. [20]

Expectant parents are often alluded to unexpected findings through the sonographers' body language and they have expressed a desire for immediate information. [21, 22] Delays in the communication of results can prompt feelings of anger and hurt and can cause distrust in providers. [16, 22] Some sonographers experience anxiety and turmoil about the decision to inform parents as they feel responsibility to the parent, but the lack of clarity around their role in the communication of unexpected findings can pose a barrier. [19]

Further complexity for sonographers in relation to communication is created by the language and terms within guidelines or publications that diverge with those considered appropriate by parents who have received a prenatal diagnosis. An example is the "banana sign" in screening guidelines, [4] which contrasts the recommendations to avoid terms that include comparisons to fruits and vegetables within consensus recommendations. [22] Even when sonographers do not deliver the initial news, they may provide follow-up scans and should maintain awareness of communication recommendations such as the avoidance of value-laden terms, assumptions, or out-dated judgements of disability. The use of baby rather than fetus, anomaly rather than abnormality and expected rather than normal are examples of language suggestions. Sonographers must be aware that some parents chose to continue their pregnancies after complex or life-limiting diagnoses. [17]

Departmental challenges

Obstetric sonography is emotionally taxing with lack of policies, recognition and support increasing work stress and the potential for burn out. [19] Funding models may not allow adequate time for the sonographer to provide evidence informed, empathetic care and to adjust between emotionally charged appointments. Department managers must ensure adequate debriefing opportunities are available to sonographers involved in difficult cases. It is imperative that sonographers are supported by their departments with protocols around the communication of findings and procedures for arranging follow-up.

The recently published '*UK consensus guidelines for the delivery of unexpected news in obstetric ultrasound: The ASCKS framework*' [22] guide UK sonographers and support departments in formulating suitable protocols. A limitation of these guidelines within the Australasian setting is the differences in the role of a sonographer in Australia and New Zealand to that in the UK, as in the UK, sonographers are practitioners who perform scans and write their own reports. Additionally, UK sonographers primarily work within the (public) National Health Service whereas private practice is common throughout Australia. This model often means when unexpected findings are encountered, Australian sonographers are hamstrung between wanting to meet the expectations of their patients as well as those of their reporting doctors and referring doctors. A nationalised guideline to help support practices in developing suitable protocols would be highly beneficial.

A limitation of this case report is its first-person narrative style of review. The case details were obtained through medical record review and recall, and the authors acknowledge the inherent bias in the first-person narrative of patient experience and the limitations of memory. This case was reported within a parallel publication directed towards mental health professionals with the focus on the psychosocial aspects of prenatal diagnosis. [23]

Conclusion

The prenatal identification of complex obstetric anomalies such as ACC present challenges for expectant parents, sonographers and ultrasound departments. The case reported highlights the way in which the combination of the limitations of imaging technologies and human factors such as

the oversight of policies, training and a mismatch between clinical language and communication guidelines potentially heightens distress experienced by parents and complicates the role of the sonographer within prenatal diagnosis.

Sonographer education for optimal language, psychosocial aspects of prenatal diagnosis, and the differences in patient pathways and support between rural and metropolitan regions will have a positive impact on both sonographers and patients as will the development of both national and departmental policies to clarify roles of sonographers and other health and medical professionals.

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Table 1. Common sonographic features associated with agenesis of the corpus callosum during a routine 19-20 week morphology scan. It should be noted, the corpus callosum does not assume its final shape until 18-20 weeks. As such, direct visualisation and confirmation of normality may be difficult before 20 weeks.

Common sonographic features of ACC at the 19-20 week morphology scan	
Direct sonographic features	**Indirect sonographic features**
Non-visualisation of the corpus callosum, or visualisation of an underdeveloped corpus callosum	Absent or atypical cavum septum pellucidum [5, 7, 8]
	Widening of the interhemispheric fissure [2]
	Atypical course of the pericallosal artery [2, 5]
	High riding or dilated third ventricle [2, 5]
	Colpocephaly/'tear-drop' shaped lateral ventricles † [2, 5, 9]
	Ventriculomegaly † [2, 5, 9]
	Three-line sign—three echogenic lines that run parallel in the upper cranium [5]

† The lateral ventricles commonly increase in size as gestation advances and may be of typical size at the morphology scan. [9]
ACC = agenesis of the corpus callosum

Table 2. Common expectant parent psychological responses and recommendations to reduce distress.

	Psychological responses	Recommendations to reduce distress
Initial	Shock [21, 22] Disorientation [18] Confusion [16, 21, 22] Denial [22] Anger [16] Fear [16] Helplessness [12] Hope [21]	Value-free language; *unexpected* rather than *abnormal* or *problem* [17, 22] Discuss the findings in a quiet, private area [22] Clear and honest communication [16, 22] Use the term *baby* rather than *fetus*, unless the parent has shown a preference for fetus [16, 22] Provide information in a variety of formats [18] about the findings and support [22] Empathise with distress, if present, rather than sympathy about the diagnosis [16, 22] Provide family centred-care [16] Sensitivity within follow up appointments [2] Ultrasounds can be special moments, take a few minutes to view the typically developed features rather than focus only on the anomalies [17]
Ongoing	Ongoing anxiety [13] Worries about delivery, care pathway and bonding [12] Hope [17] Adjustment [13] Fear of judgement about decisions [13] Post-traumatic stress disorder [14, 15]	

Figure 1:

A transvaginal ultrasound of the fetal head at 31 weeks and 0 days showing ventriculomegaly with the lateral ventricle measuring 19mm as demonstrated by the callipers.

Figure 2:

A fetal MRI scan performed at 33 weeks and 6 days gestation showing colpocephalic configuration of ventricles (B), severe commissural dysgenesis (C) due to complete agenesis of the corpus callosum (As indicated by the white arrows in the coronal (C) and sagittal (D) planes). A tiny residual anterior commissure was present although not represented in these images.

Figure 3:

An ultrasound of the fetal head at 37 weeks and two days. The white arrows indicate bilateral ventriculomegaly (A) and an absence of the cavum septum pellucidum (CSP) (B). It should be noted that in typically developing fetuses, the CSP may not be seen late in the third trimester.

Figure 4:

MRI images performed on the infant at three months of age confirming the absence of the corpus callosum in the coronal (A) and sagittal (B) planes. The corpus callosum should be seen in the regions of the white arrows but in this case, is absent.