Kentucky Anabaptists' Knowledge of Newborn Screening[1]

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Abstract: The Plain Anabaptist population in Kentucky is growing, with the Amish population alone expected to double about every 20 years. Anabaptists in Kentucky are largely composed of Amish (both Old Order and New Order) and Old Order and Conservative Mennonites. All Anabaptists experience an increased incidence of recessive genetic disorders. Newborn screening (NBS) is a state-regulated program that identifies inherited and congenital disorders and conditions. The purpose of this study is to determine the knowledge of Kentucky Amish and Mennonite communities regarding NBS. A researcher-adapted questionnaire was mailed to Kentucky Amish and Mennonite households, and 292 respondents returned the questionnaire. The majority (85.2%) reported at least "a little" knowledge of NBS; however, only 6.8% reported "a lot" of knowledge. Respondents identified the midwife as the most common source of information related to NBS; however, 18% reported NBS was not offered by any provider. There was a statistically significant relationship between knowledge of NBS and children receiving NBS, indicating that knowledge of NBS increased participation in NBS. As the midwife is the most common source of NBS knowledge, an educational program for midwives as well as the Anabaptist communities throughout Kentucky is planned.

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Kentucky is one of the few southern states with a significant Anabaptist population, including Old Order Amish, Old Order Mennonites, Conservative Mennonites, New Order Amish, and others (Donnermeyer & Luthy, 2013). The Anabaptist population in Kentucky is growing rapidly. From 2000 to 2012, the number of Amish settlements in Kentucky increased from 22 to 48 and the population grew from 4,850 to 14,215 (Young Center for Anabaptist and Pietist Studies, 2021). It is estimated that the Amish population will double every 20 years (Donnermeyer, 2021). Hart County is the most densely populated Amish (both Old Order and New Order) community in Kentucky, comprising 5.9% of the county population (J. Donnermeyer, personal communication, September 27, 2021). The precise number of the other Anabaptist groups in Hart County is not known but likely raises the proportion to over 7.0%.

The Anabaptist communities have been described as both "separatist and community-oriented," referring to those outside of their tradition as "English" (Kraybill et al., 2013). Due to the small number of founders and the practice of marrying within their religious community, the Anabaptists experience an increased incidence of recessive genetic disorders such as maple syrup urine disease (MSUD) and glutaric acidemia type 1 (GA1) (Lopes et al., 2016). Hunt et al. (2018) found that more than one-third of Kentucky Anabaptist households reported a family member with a diagnosed or suspected genetic condition.

Newborn Screening (NBS) is critical for the identification of potentially lethal or debilitating diseases and disorders that are not apparent at the time of birth (Newborn Screening Technical Assistance and Evaluation Program, 2021). Anabaptists have a keen sense of community and responsibility for the care of family and a preference to receive care in the home rather than at a hospital. Most Anabaptist families choose an in-home birth with a midwife,[2] precluding mandatory newborn screening in the hospital (Anderson & Potts, 2020). However, there is a gap in the literature regarding knowledge of Newborn Screening (NBS) on the part of Kentucky Anabaptists. The purpose of this study is to determine the level and source of knowledge of Kentucky Amish and Mennonite communities regarding NBS. The research questions include:

1. What is the self-reported knowledge regarding NBS of Kentucky Amish and Mennonite households?
2. What is the source of Kentucky Amish and Mennonite households' knowledge of NBS?
3. How does Kentucky Amish and Mennonite self-reported knowledge of NBS relate to NBS rates?

Background

In the eighteenth century, religious persecution led to the migration of Anabaptist groups such as the Amish and Mennonites from Europe to Pennsylvania (Nolt, 2015). Small groups of Anabaptists

[2] Most Anabaptists use traditional or lay midwives. Lay midwives are community midwives with practical experience such as an apprenticeship with a practicing midwife; however, they are uncertified or unlicensed with the state.

have since migrated to other states, including Kentucky. Migration of Anabaptist families to Kentucky began in 1958 with the first settlement near Guthrie in Todd County (Donnermeyer & Anderson, 2014).

Genetic drift is a lack of genetic diversity, and it facilitates the loss of some traits as well as the increased rate of rare genetic traits and diseases (Lopes et al, 2016). The *founder effect* is the type of genetic drift seen in the Anabaptist population (Brennan, 2018). Due to the small number of founders and the practice of endogamy (marriage within one's religious group), the Anabaptists experience a fair degree of genetic isolation (Strauss et al., 2012). This isolation leads to an increase in the incidence of recessive genetic disorders when compared to the general population (Lopes et al., 2016; Sieren et al., 2016).

Genetics Clinics

In 1989, the Clinic for Special Children was established in Strasburg, Pennsylvania, by Dr. D. Holmes Morton, his wife, Caroline, and local Anabaptist residents (Hendricks, 1994). The initial goal of the clinic was prevention of disability and death of children with MSUD and GA1 through early diagnosis and intervention (Morton et al., 2003). Morton's success in diagnosing and treating MSUD and GA1 in Anabaptist children expanded the knowledge and treatment of these genetic conditions, which improved quality of life.

MSUD, a recessive inherited disease, is noted in the Old Order Mennonite community. It is a rare metabolic disorder in which symptoms begin to emerge within 48 hours of birth and, if untreated, causes progressive brain damage that can lead to death within weeks or months (National Organization for Rare Disorders, 2020). According to Frazier et al. (2014) the worldwide incidence of MSUD is ~1 in 185,000 births while the incidence in Old Order Mennonites is ~1 in 200 births. NBS provides early detection, allowing for early intervention with nutritional therapy to promote normal growth and development (Frazier et al., 2014).

GA1 is a genetic disorder that is more commonly seen in the Anabaptist population and especially in the Old Order Amish, with a birth incidence as high as ~1 per 400 births, while the worldwide birth incidence is ~1 per 90,000 (Strauss et al., 2020). GA1 is a metabolic disorder that causes an accumulation of metabolites in the brain, leading to permanent brain damage resulting in movement disorders. A study by Strauss et al. (2020) indicates that NBS in combination with "appropriate metabolic formula and timely intravenous (IV) infusion therapy during the first two years of life, prevents more than 90% striatal injuries while supporting normal growth and psychomotor development" (p. 326).

Following Morton's initial work, similar clinics serving the Plain Anabaptists were established in Ohio and Indiana. The initial meeting to discuss the establishment of a genetic clinic in western Kentucky occurred in March of 2009. During a visit to Christian County in May 2015, Dr. Morton provided guidance in the creation of a Kentucky genetic clinic (Grace, 2017). Hunt et al. (2018) completed a needs assessment that demonstrated necessity for a clinic in the central and western areas of Kentucky. The WeCare Clinic-Medical Care for Special Needs (WeCare Clinic) in

Pembroke, Kentucky, opened in December of 2020 to serve the needs of the Amish, Mennonite, and English communities in central and western Kentucky (WeCare Clinic, 2020).

Newborn Screening

NBS is essential in the detection of inherited and congenital disorders and conditions that, if untreated, could lead to disability or death (National Library of Medicine, 2021). NBS is composed of blood tests for inherited disorders, hearing assessment, and pulse oximetry to screen for congenital heart defects. NBS is a state-run public health program, with each state adopting a panel of tests and regulations to manage testing and results (Kelly et al., 2016). Efforts to screen newborns began in the 1960s with the implementation of the phenylketonuria (PKU) screening test (Baby's First Test, 2018). The Secretary of the Department of Health and Human Services (HHS) and the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommend a standardized list of disorders to be included in the state-run newborn screening program (ACHDNC, 2020; Kelly et al., 2016). This list is known as the Recommended Uniform Screening Panel (RUSP) (Health Resources & Services Administration, 2020). Over 12,000 newborns with genetic diseases or disorders are identified from NBS each year in the United States (Newborn Screening Technical Assistance and Evaluation Program, 2020).

All U.S. states require testing for at least 30 disorders (NICHD, 2021). The Kentucky Cabinet for Health and Family Services (CHFS) operates the Kentucky Newborn Screening Program and specifies 54 disorders for screening (Newborn Screening Program, 2021). Kentucky law requires Krabbe disease to be included with the RUSP disorders. All newborns in Kentucky are required to receive NBS (Baby's First Test, 2021). The James William Lazzaro and Madison Leigh Heflin Newborn Screening Act (1966/2020) mandates infants less than 28 days of age be screened for inherited and congenital abnormalities prior to discharge from the hospital or institution. The one exception from screening is if the child's parents "are members of a nationally recognized and established church or religious denomination, the teachings of which are opposed to medical tests, and who object in writing to the testing of his or her child on that ground" (James William Lazzaro and Madison Leigh Heflin Newborn Screening Act, 1966/2020, sec. 5).

The cost of NBS in Kentucky is $150 and is the responsibility of the newborn's "responsible party" (Baby's First Test, 2021). The state does not cover the cost of the test if the newborn does not have insurance. The test is charged to the *submitter*, who in turn bills the uninsured patient (Baby's First Test, 2021). The submitter is defined as a hospital, primary care provider, health department, birthing center, laboratory, or midwife, and it is the submitter's responsibility for the original testing (Newborn Screening Program, 2021). Plain Anabaptists do not participate in government or commercial insurance programs and pay directly for medical services (Anderson & Potts, 2020). Home births are preferred, with the services of a midwife. Newborn screening is completed outside the hospital setting if the family chooses and the attending midwife will facilitate testing (J. Schmucker, personal communication, August 11, 2021).

Method

The present study is a descriptive design utilizing a convenience sample of Amish and Mennonite households in Kentucky. Permission was obtained from Christine M. Seroogy, professor in the School of Medicine and Public Health at the University of Wisconsin-Madison, to utilize and revise the survey from the study "Cross-Sectional Survey on Newborn Screening in Wisconsin Amish and Mennonite Communities" (Sieren et al., 2016). The final questionnaire was modified with input from Amish and Mennonite insiders and consisted of 17 questions including yes or no, fill in the blank, Likert scale, and multiple choice. Demographic information was obtained by asking these questions: "What group are you a member of? (New Order Amish, Old Order Amish, Old Order Mennonite, Conservative Mennonite, or Other)"; "Do you have children?" "If yes, how many?"; "How many people live in your household?"; and "What is the name of the county in which you live?"[3]

Approval was obtained from a university institutional review board. Completion and return of the questionnaire were considered implied consent.

Procedure

The questionnaire, a letter from the WeCare Clinic chairman of the board, and an informed consent document were mailed to the 1,850 households on the mailing list maintained by the WeCare Clinic. Participants were asked to return the questionnaires to an anonymous box at the WeCare Clinic or to use the enclosed postage-paid envelope. A reminder to complete the questionnaire appeared in the newsletter. Expenses associated with printing and mailing were funded by the WeCare Clinic Board; Sigma Theta Tau International, Kappa Theta chapter; and the researchers. The data analysis was generated using SPSS software, version 27. Data was analyzed utilizing descriptive and inferential statistics.

Variables

NBS was defined on the questionnaire as being "a series of tests (blood, hearing, and heart) that can detect treatable diseases in a newborn baby." The concept of knowledge of NBS was measured by the question "How much do you know about newborn screening?" The responses on a Likert scale ranged from 1 ("a lot") to 5 ("never heard of it"). The source of NBS knowledge was measured by the question "If you knew about one or more of these screenings, how did you learn about it (circle all that apply)?" The respondent choices included "midwife," "brochure," "family member," "community meeting," "another source," and "never heard of it."

Results

Sample Demographics

Questionnaires were returned from 292 Kentucky Anabaptist households, for an approximate 16% rate of return, compared to a rate of about 25% from the original Wisconsin study (Sieren et al.,

[3] Interested parties may contact the first author for a copy of the questionnaire used in this study.

2016). The largest percentage of respondents were Old Order Amish at 41.1%, and nearly all (96.2%) reported having children. See Table 1 for all sample demographics. Two-thirds of the 292 respondents live in six counties, as shown in Figure 1.

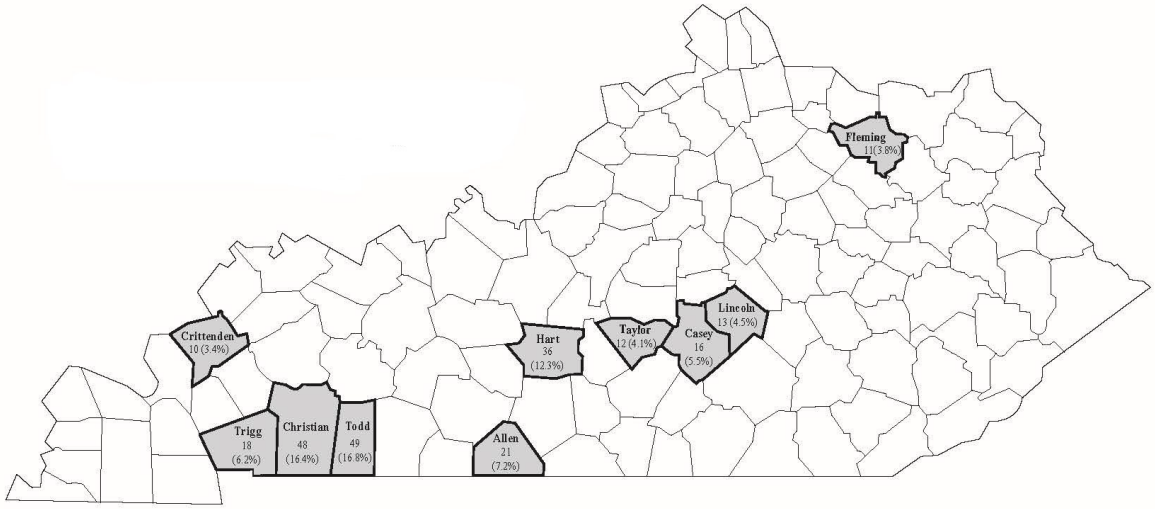
Table 1

Sample Demographics (N = 292)

	n (%)	Mean (*SD*)
Anabaptist group		
Old Order Amish	120 (41.1)	
Old Order Mennonite	105 (36.0)	
Conservative Mennonite	52 (17.8)	
Other	11 (3.4)	
Reported having children	281 (96.2)	
Total number of children		6.5 (3.1)
Live in household		6.9 (3.0)

Figure 1

Kentucky Counties with 10 or More Respondents



Findings

About 85.2% of respondents had at least "a little knowledge" of NBS, while 12.7 % had "no knowledge." Two percent of the respondents did not respond to the question. Only 6.8% respondents reported that they had "a lot of knowledge." Several respondents asked if NBS is the same as the PKU test, and several wondered what comprises assessment of the heart. A majority (79.9%) of the households reported that their children received some newborn screening tests. Nearly 56% of the responding households reported that all the children of the family received

NBS. A Pearson chi-square test of independence was performed to evaluate the relationship between knowledge of NBS and household children receiving NBS. This association was found to be significant, $x^2(1, N = 268) = 70.54, p = <.001$. The respondents with knowledge of NBS were more likely to have their children receive NBS. In fact, 100% of the children of those households who reported a lot of knowledge received NBS.

Respondents reported learning about newborn screenings from a variety of sources, including midwife (142), family member (66), brochure (55), community meeting (17), and other sources (73). As previously mentioned, Plain Anabaptist families prefer home births with a midwife present, and in this study, 1,109 (62.5%) children were born in the home and 665 (37.5%) were born in a hospital.

Respondents gave a variety of reasons for not participating in NBS. This question allowed multiple responses, and 56 reported that they "did not know about NBS" and 52 reported that "NBS was not offered by health care provider." Fourteen reported that travel for testing was too difficult, and eight said that NBS was too expensive.

Discussion

The purpose of this study was to determine the knowledge of Kentucky Amish and Mennonites regarding NBS. The majority of respondents reported "some" knowledge of NBS; however, only a small number reported "a lot" of knowledge. A previous study by Sieren et al. (2016) noted that the majority of Wisconsin Amish and Mennonites reported "some" or "a lot" of knowledge regarding NBS. Further indications of respondents' knowledge level included their question if NBS is the same as the PKU test and their belief that they did not need NBS if they have no family history of genetic conditions or if the newborn appears healthy.

Additional research utilizing focus groups and key informant interviews is needed to determine the details of Plain Anabaptist knowledge and possible objections to newborn screening tests. Focus groups and key informant interviews will allow the researcher to answer questions related to what is known and why it is important.

The most common source of information regarding NBS was a midwife. This is consistent with the findings of Sieren et al. (2016), who found that the majority of Wisconsin Amish and Mennonite communities reported that their source of NBS information was a midwife. Although a midwife was reported as the most common source of information regarding NBS in the current study, several respondents reported that NBS was not offered by their provider. It is also noted that the majority of Anabaptist children in this study were born at home, where childbirth is overseen by the midwife and NBS is not required by law. All children born to parents who self-reported *a lot* of knowledge regarding NBS received the screening, indicating that education of Anabaptist parents may improve NBS rates.

The support of the WeCare Clinic board of directors and staff potentially improved the survey response rates. These results are not generalizable to all Plain Anabaptists, as the survey was only sent to households included in the WeCare Clinic mailing list, and a vast majority of the respondents resided in Kentucky.

These study results demonstrate a need for additional community education regarding NBS. Families would benefit from education on the purpose of NBS and its components. The study also revealed that most of the respondents received NBS information from their midwife. Hence, the education of midwives regarding the importance of NBS is essential. Development and implementation of an educational program for midwives is planned as well as an informational pamphlet to be placed at the WeCare Clinic and included in the statewide newsletter. Future studies to evaluate the effectiveness of this intervention would be beneficial for other Plain Anabaptist communities.

Access to care and testing is a critical issue. The WeCare Clinic was established for the community to care for those with genetic disease and disability. Additional future research should include determining which Anabaptists utilize the WeCare Clinic and NBS. Travel distance is a deterrent to care. As the clinic is located in the Pennyrile (western) region of Kentucky, do the Anabaptists in distant areas of the state utilize the clinic? Are they hesitant to have newborn screening performed because the genetic clinic is not accessible due to travel?

Conclusion

Timely NBS and intervention for affected newborns is paramount for the Plain Anabaptist community due to the increased risk of recessive genetic disease. Many Anabaptists are unaware of the risk and are hesitant to "look for trouble." Additional research and interventions to educate Anabaptists about the risk of not receiving NBS and the availability of resources would benefit all Anabaptists.

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