The Down Syndrome Diagnostic Experience: Support Beyond Labor, Delivery, and Diagnosis

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BACKGROUND & AIMS:

Mothers often report negative experiences with maternal healthcare both prenatally and postnatally. Similar patterns have been observed among mothers of children with Down syndrome (DS), particularly surrounding the delivery of the DS diagnosis and the type and perceived quality of support provided. Despite these reports, few studies have systematically examined contributors to these experiences, such as perceived discrimination, medical trauma, inadequate support, or identified ways clinicians can foster a more affirming, culturally responsive diagnostic environment during this critical period.

This study aimed to examine the type and perceived quality of support provided to mothers of color during their child's DS diagnosis, both pre- and postnatally, with the following questions:

1. What differences may exist in medical care experiences for families of color?
2. What types of resources, if any, were provided to families at the time of diagnosis?
3. How much perceived support were families provided with from their birth teams (social worker, obstetrician, pediatrician, nurse, etc.) during the diagnostic process?

METHODS AND MEASURES:

Participants

- Participants were mothers who had given birth to a child with Down syndrome.
- *N* = 13 completed responses, ages 18-33 (35.7%) 34-48 (64.3%)
- Race/Ethnicity: (n=4) White 35.7%, (n=8) Black 57.1% , (n=1) Multi-racial, (n=1) Hispanic 7.1%
- Income: 25-50k 7.1%, 50-75k 14.3%, 75-100k 50% , 100-135k 7.1%, 150-175k 14.3%, more than 200k 7.1%
- Participants completed multiple choice questions on an online survey, protocol, assessing:
  - Perceived discrimination using the PhenX Toolkit Discrimination in Health Care,
  - Medical trauma experiences, and
  - Nature of support provided.
- Each item (e.g., My birth team (doctors, nurses, social workers) at the time of diagnosis, provided me with the physical/emotional support I needed.) was rated using a 5-point Likert-type scale (1 = strongly agree, 5 = strongly disagree).
- Participants also completed open ended questions, which allowed them to further describe their diagnostic experience and the type of support they would have liked to receive from their birth team.

Resources for extended family members.

Quote from participant: *"I am a single parent with no support. financially we do find. but my child with DS is missing the family contact. plus the family i do have shy away from her. I feel extended family need additional support or different type of information to support a diagnosis."*

Begin diagnosis delivery with strengths and local resources tailored to the mother.

Quote from participant: *"What bothered me the most was their need to tell me every possible thing that could go wrong with my child. Every health issue that my baby could possibly have or get. My thoughts as a mother of three already, was that the doctors didn't tell me everything that could go wrong or possible health issue that my other children could have had…"*

Have caution with language recommending/hinting termination.

Quote from participant: *"options....the word options is a polite way of saying you can abort if you want to. I hate that word. do not use the word option. it's not a choice. its a personal decision that everyone know they have…"*

Be mindful of tone and body language.

Quote from participant: *"Doctors and teams need to be aware that their tone and body language confirm our fears or give us hope."*

RESULTS:

- 65% of mothers reported receiving their child's diagnosis prenatally.
- 40% of mothers reported receiving a recommendation to terminate the pregnancy.
- Approximately half (50%) of mothers reported feeling that they had the emotional and physical support they needed at the time of diagnosis.
- Across the full sample, mothers most commonly reported receiving informational pamphlets and specialist referrals.

Discussion:

In this sample, most mothers received a prenatal diagnosis of Down syndrome; however, prenatal identification of their loved ones disability was not coupled with increased access to supportive resources. Many mothers also reported receiving recommendations to terminate, which may reflect directive practices rather than supportive, family-centered discussions. Such recommendations seem to significantly influence parental perceptions of the diagnosis and shape early decision-making during an already vulnerable period. Even though about half of mothers reported that their birth team met their emotional and physical support needs, their qualitative narratives often described more complex or negative experiences, including limited resources and recommendations to terminate. This suggests that broad ratings of "support" may not fully capture the nuances of how diagnostic encounters are experienced in real time.

LIMITATIONS:

The sample size of this study was fairly small which limited our ability to detect significant differences. This study is ongoing.

Future Implications:

Future implications include a larger, mixed-methods study and the development of culturally responsive clinician guidelines for delivering Down syndrome diagnoses. Incorporating routine patient-reported feedback may improve understanding of families' lived experiences beyond initial clinical encounters.

Conclusion:

These preliminary results demonstrate potential disparities in diagnosis delivery and post-diagnostic support for mothers of color following a Down syndrome diagnosis. Despite receiving prenatal diagnoses, mothers of color reported fewer resources and mixed perceptions of support, suggesting gaps in equitable, patient-centered care.