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| **About the Measure** | |
| **Domain:** | Sickle Cell Disease Pregnancy |
| **Measure:** | Invasive Prenatal Diagnosis |
| **Definition:** | Invasive procedures for prenatal diagnosis include amniocentesis and chorionic villus sampling. |
| **Purpose:** | Invasive prenatal diagnosis procedures are well-established methods to test for chromosomal and genetic abnormalities (e.g., Down syndrome, sickle cell disease) prior to birth. |
| **Essential PhenX Measures:** | NA |
| **Related PhenX Measures:** | NA |
| **Measure Release Date:** | TK Team to add |

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| **About the Protocol** | |
| **Protocol Release Date:** | TK Team to add |
| **PhenX Protocol Name:** | Invasive Prenatal Testing Guidelines |
| **Keywords:** | NA |
| **Protocol Name from Source:** | International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) Practice Guidelines: Invasive procedures for prenatal diagnosis |
| **Description:** | This protocol summarizes the 2016 recommendations from the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) for performing amniocentesis and chorionic villus sampling. |
| **Specific Instructions:** | None |
| **Protocol:** | **Invasive Prenatal Testing Guidelines**  The 2016 recommendations by the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG; see Source section) summarize considerations for performing amniocentesis, chorionic villus sampling, and fetal blood sampling. The guidelines provide a definition of each procedure and cover considerations for performing the techniques, timing, laboratory aspects, potential complications, eligibility, and types of genetic testing. Additionally, the guidelines indicate level of evidence and grade of recommendation based on the available literature. |
| **Selection Rationale:** | The PhenX Sickle Cell Disease Pregnancy Working Group (WG) selected the 2016 recommendations from the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) as the gold standard for performing invasive prenatal diagnostic testing. The WG recommended it be included in Supplemental Information due to investigator burden. |
| **Source:** | Ghi, T., Sotiriadis, A., Calda, P., Da Silva Costa, F., Raine-Fenning, N., Alfirevic, Z., & McGillivray, G.; International Society of Ultrasound in Obstetrics and Gynecology (ISUOG). (2016). ISUOG Practice Guidelines: Invasive procedures for prenatal diagnosis. *Ultrasound in Obstetrics and Gynecology : The Official Journal of the International Society of Ultrasound in Obstetrics and Gynecology*, *48*(2), 256–268. <https://doi.org/10.1002/uog.15945> |
| **Availability:** | Available |
| **Life Stage:** | Adolescent, adult |
| **Language:** | English |
| **Participant:** | Pregnant women with sickle cell disease |
| **Personnel and Training Required:** | See Ghi et al. (2016) for details. |
| **Equipment Needs:** | See Ghi et al. (2016) for details. |
| **General References:** | Hanson, B., Scotchman, E., Chitty, L. S., & Chandler, N. J. (2022). Non-invasive prenatal diagnosis (NIPD): How analysis of cell-free DNA in maternal plasma has changed prenatal diagnosis for monogenic disorders. *Clinical Science (London, England : 1979)*, *136*(22), 1615–1629. <https://doi.org/10.1042/CS20210380>  Cutts, A., Vavoulis, D. V., Petrou, M., Smith, F., Clark, B., Henderson, S., & Schuh, A. (2019). A method for noninvasive prenatal diagnosis of monogenic autosomal recessive disorders. *Blood*, *134*(14), 1190–1193. <https://doi.org/10.1182/blood.2019002099>  Salomon, L.J., Sotiriadis, A., Wulff, C.B., Odibo, A., & Akolekar, A. (2019). Risk of miscarriage following amniocentesis or chorionic villus sampling: systematic review of literature and updated meta-analysis. *Ultrasound Obstet Gynecol*, 54:442-451. <https://doi.org/10.1002/uog.20353> |
| **Mode of Administration:** | Clinical examination |
| **Derived Variables:** | None |
| **Requirements:** | |  |  | | --- | --- | | **Requirements Category** | **Required (Yes/No):** | | Major equipment | Yes | | Specialized training | Yes | | Specialized requirements for biospecimen collection | Yes | | Average time of greater than 15 minutes in an unaffected individual | Yes | |
| **Annotations for Specific Conditions:** | None |
| **Process and Review:** | NA |