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| **Domain:** | Reproductive Health |
| **Measure:** | Male Reproductive Tract Birth Defects |
| **Definition:** | Questions to determine whether a male participant has ever had certain conditions that could have developed in utero. Cryptorchidism is failure of one or both of the testes to descend into the scrotum. Hypospadius is a developmental anomaly in which the male urethra opens on the underside of the penis or on the perineum. |
| **Purpose:** | The purpose of these questions is to determine whether the participant had ever had the following abnormalities that developed in utero: Cryptorchidism, Hypospadius, and/or other related conditions. These conditions are suspected of being hereditary and/or influenced by environmental factors, may reflect abnormalities of androgen production, and may be passed to offspring. |
| **Essential PhenX Measures:** | Current Age Gender |
| **Related PhenX Measures:** |  |
| **Collections:** | Cancer-Related Medical History Congenital Defects Infertility and Sexual Dysfunction Infant Complications |
| **Keywords:** | Reproductive health, male reproductive tract birth defects, male in utero conditions, Cryptorchidism, Hypospadius, penis, testicles, urinary tract |

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| **Protocol Release Date:** | February 26, 2010 |
| **PhenX Protocol Name:** | Male Reproductive Tract Birth Defects |
| **Protocol Name from Source:** | The Expert Review Panel has not reviewed this measure yet. |
| **Description:** | The male participant answers whether he was ever diagnosed with Cryptorchidism, Hypospadius and/or other related conditions. |
| **Specific Instructions:** | The questions were originally asked of partners of pregnant women but the WG recommends asking them of adult males.  Note: Cryptorchidism is failure of one or both of the testes to descend into the scrotum. Hypospadius is a developmental anomaly in which the male urethra opens on the underside of the penis or on the perineum. |
| **Protocol:** | 1. Were you born with one or both of your testicles undescended (not completely down in the scrotum)?  [ ] 0 No (go to 5)  [ ] 1 Yes  [ ] 2 Don't know (go to 5)  2. If yes, which testicle was this?  [ ] a Right  [ ] b Left  [ ] c Both  3. Did the testicle go down to the scrotum by itself?  [ ] 0 No  [ ] 1 Yes  [ ] 2 Don't know  4. Did you receive treatment?  [ ] 0 No  [ ] 1 Yes  [ ] 2 Don't know  5. What treatment?  [ ] Surgery  [ ] Hormones  [ ] Other (specify)  [ ] Don't know  6. Has your doctor or another health care provider ever told you that you had other diseases of the penis, testicles, urinary tract or scrotum (specify)?  \_\_\_\_\_\_\_\_\_\_\_  7. Has your doctor or another health care provider ever told you that you had hypospadius?  [ ] No  [ ] Yes  8. What was your most recent treatment or medication (if any)?  [ ] Specify  [ ] Don't Know  [ ] Year |
| **Selection Rationale:** | The presence of these physical conditions is often associated with abnormalities of androgen production such as Kallmann's Syndrome and 5 alpha reductase deficiency, and may be associated with certain chemical exposures in utero. The Study for Future Families' protocol was selected because it specifically addresses physical exam findings. |
| **Source:** | Swan SH, Brazil C, Drobnis EZ, et al. "Geographic Differences in Semen Quality of Fertile U.S. Males." Environmental Health Perspectives 111(4): 414-420.  Utilizes questionnaire generated by the Study For Future Families |
| **Life Stage:** | Adult |
| **Language of source:** | English |
| **Participant:** | Males 18-53 |
| **Personnel and Training Required:** | None |
| **Equipment Needs:** | Paper and pencil |
| **Standards:** | |  |  |  |  | | --- | --- | --- | --- | | **Standard** | **Name** | **ID** | **Source** | | Common Data Element (CDE) | Male Reproductive System Birth Defect | 3007471 | [CDE Browser](https://cdebrowser.nci.nih.gov/CDEBrowser/search?elementDetails=9&FirstTimer=0&PageId=ElementDetailsGroup&publicId=3007471&version=1.0) | | Logical Observation Identifiers Names and Codes (LOINC) | Male reproductive birth defects proto | 62664-8 | [LOINC](http://s.details.loinc.org/LOINC/62664-8.html?sections=Web) | |
| **General references:** | Swan SH, Main KM, Liu F, Stewart SL, Kruse RL, Calafat AM, et al; Study for Future Families Research Team. (2005). Decrease in anogenital distance among male infants with prenatal phthalate exposure. Environ Health Perspect., 113(8):1056-61. Erratum in: Environ Health Perspect. 2005 Sep;113(9):A583. PubMed PMID: 16079079; PubMed Central PMCID: PMC1280349. |
| **Mode of Administration:** | Self-administered questionnaire |
| **Derived Variables:** |  |
| **Requirements:** | |  |  | | --- | --- | | **Requirement Category** | **Required** | | Major equipment | No | | Specialized training | No | | Specialized requirements for biospecimen collection | No | | Average time of greater than 15 minutes in an unaffected individual | No | |
| **Process and Review:** | The Expert Review Panel has not reviewed this measure yet. |