Study on the Effect of Socio-Demographic Factors on Different Congenital Disorders

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Abstract

Congenital disorders define the disease that occurs since the birth of a baby. Down syndrome, Turner syndrome, cleft lip, and congenital heart disease are the most common congenital disorders worldwide. A retrospective study was carried out, examining the effect of sociodemographic factors on congenital anomalies in the state of West Bengal, India, over a period of 6 years. A total of 595 cases with congenital disorders including Down syndrome, Turner syndrome, and other abnormalities (cleft lip/palate, syndactyly, ambiguous genitalia) were statistically analyzed along with the sociodemographic characteristics through Statistical Analysis System (SAS) 9.3.2. Down syndrome is seemed to be associated with age, ethnicity, parental addiction, especially smoking, while Turner syndrome is associated with ethnicity and gender. Other congenital disorders such as ambiguous genitalia are found to be associated with maternal addiction.

Keywords: congenital disorders, down syndrome, turner syndrome, cleft lip/palate, syndactyly, ambiguous genitalia, sociodemographic factors

1. Introduction

Congenital disorder, which is a health hazard since birth, may be caused mostly by genetic anomalies [1]. Some congenital disorders are hereditary that are transmitted through parents to the children [2]. Several types of congenital disorders are present of which the most common congenital disorders are Down syndrome, Turner syndrome, congenital heart diseases, etc., are considered the most common and severe disorders since birth [3–5]. This type of disorder cannot be cured but managed, though some of them can be prevented or cured such as cleft lip/palate through surgical intervention [6]. The exact cause of congenital abnormalities is not fully understood. Sometimes it depends on genetic or infectious factors, and sometimes it may be caused by nutritional or environmental factors [7–9].

In this book chapter, we have discussed the possible effect of sociodemographic factors, including environmental and behavioral facets on congenital disorders [10]. The main focused congenital disorders are Down syndrome (2n = 47, XX/XY, +21) and Turner syndrome (2n = 45, X). Down syndrome is a genetic condition with an extra chromosome (chromosome no. 21) that presents since birth and this condition results in developmental delay along with associated diseases such as heart disease, intestinal obstruction [11–13]. This “package” of the 21st chromosome (trisomy or
three copies of chromosome 21) is caused due to nondisjunction of chromosome 21 in meiotic cell division during the development of the sperm cell or the egg cell [14]. Studies suggest that the advanced maternal age, the addiction of the mother as well as the father may be the prime cause for this kind of condition to their child [15, 16]. However, sociodemographic factors are also thought to be associated with these diseases [17]. Another common congenital disorder is Turner syndrome, which is also discussed in this chapter. Turner syndrome only affects females and one of the X chromosomes (sex chromosome) is fully or partially missing [18]. This condition results in a variety of medical and developmental problems such as short stature, webbed neck, delayed development of ovaries, heart defects, loss of puberty and menstruation, infertility [19]. Most of the cases of Turner syndrome cannot be cured, though hormone therapy can be useful for treatment in some cases [20]. Turner syndrome occurs due to the nondisjunction of the X chromosome in meiotic cell division during the formation of an egg or sperm cell in a parent (prior to conception) [21]. The other discussed congenital anomalies include cleft lip/palate, syndactyly, and ambiguous genitalia. Cleft lip/palate is a common birth condition. It occurs alone or as part of a genetic condition or syndrome [22, 23]. Symptoms arise from the opening in the mouth and include the difficulty in speaking and feeding [24]. Surgeries are the useful treatment for this condition [25]. Sometimes speech therapy helps to improve the speaking ability [26]. Syndactyly is the fusion of the bone or skin in the hand or foot digits [27]. This condition is due to developmental anomalies. Ambiguous genitalia is a rare condition in which an infant's external genitals do not appear to be clearly manifested as a either male or female [28]. In a baby with ambiguous external genitalia, the genitals may be incompletely developed or the baby may have characteristics of both sexes [29]. Karyotype helps in determining the proper sex of the patients and subsequent surgical intervention is required to cure the affected individuals.

The sociodemographic features involve a combination of social and demographic facets. Social facets include behavioral factors such as addiction where the demographic part includes age, gender, race, etc. [30]. This work is a descriptive analysis of all different sociodemographic factors, including other diseases, associated with studied congenital disorders.

2. Materials and methods

Data were collected from a retrospective study, examining the sociodemographic factors along with a few behavioral characteristics from the state of West Bengal, India, along with the diagnostic information about common congenital disorders for the 595 samples over a period of 6 years (2011–2017). Patients were diagnosed at the Centre for Genetic Studies, Maulana Abul Kalam Azad University of Technology. All data were recorded after taking the informed consent from the participants. Collected data were entered using a database management software MySQL. Entered data were exported to SAS (Statistical Analysis Software version 9.3.2) and analyzed for understanding the patterns and predictors of the identified genetic disorders. Descriptive analyses were conducted to determine the frequency and proportion (along with corresponding 95% confidence intervals and p values to denote whether the categories for each factor had a statistically significant different distribution of the proportions) of the sociodemographic factors (gender, religion), behavioral factors (consanguinity, contraception use, addiction), clinical history (history of spontaneous abortion, diabetes, hormonal deficiency), family history (history of congenital abnormalities among relatives and disease distribution if any such as Down, Turner, and other congenital
abnormalities) among the sampled population. The sum of the total frequencies in all the categories in each variable will not be equal to 595 as there were multiple missing values for different variables and while analyzing the distribution and associations, they were dropped. Binary and multinomial, and logistic regressions were next conducted to determine the association (odds ratios, corresponding 95% confidence intervals, and p values) between the study variables and diagnosed diseases. Multiple logistic regressions to determine the association between the variables adjusted for all others could not be done for inadequate sample size. The results of the analyses are presented in Tables 1–9. Each table is followed immediately by the interpretation of the observed results presented in each of these tables, respectively.

| Variables                                             | Categories            | N   | 95% CI               | P value |
|-------------------------------------------------------|-----------------------|-----|----------------------|---------|
| Gender                                                | Male                  | 279 | 46.89 (42.87–50.91)  | <.0001  |
|                                                       | Female                | 313 | 52.61 (48.58–56.63)  |         |
| Religion                                              | Muslim                | 152 | 28.52 (24.67–32.36)  | <.0001  |
|                                                       | Hindu                 | 381 | 71.48 (67.64–75.33)  |         |
| History of consanguinity                              | Yes                   | 28  | 4.71 (3.00–6.41)     | <.0001  |
|                                                       | No                    | 567 | 95.29 (93.59–97.00)  |         |
| Contraceptives used                                   | Yes                   | 104 | 17.48 (14.42–20.54)  | <.0001  |
|                                                       | No                    | 491 | 82.52 (79.46–85.58)  |         |
| Addiction of father                                   | None                  | 341 | 57.31 (53.33–61.30)  | <.0001  |
|                                                       | Smoking               | 178 | 29.92 (26.23–33.61)  |         |
|                                                       | Smoking/drug          | 65  | 10.92 (8.41–13.44)   |         |
|                                                       | Smoking/drug/alcohol  | 11  | 1.85 (0.76–2.93)     |         |
| History of spontaneous abortion                       | Yes                   | 206 | 86.19 (81.79–90.60)  | <.0001  |
|                                                       | No                    | 33  | 13.81 (9.40–18.21)   |         |
| Presence of diabetes                                  | Yes                   | 43  | 7.23 (5.14–9.31)     | <.0001  |
|                                                       | No                    | 552 | 92.77 (90.69–94.86)  |         |
| Presence of hormonal deficiencies (FSH/TSH/etc.)     | Yes                   | 62  | 10.42 (7.96–12.88)   | <.0001  |
|                                                       | No                    | 533 | 89.58 (87.12–92.04)  |         |
| History of congenital disease among first degree relatives | Yes               | 71  | 11.93 (9.32–14.55)   | <.0001  |
|                                                       | No                    | 524 | 88.07 (85.46–90.68)  |         |
| Any genetic abnormality detected                      | No                    | 308 | 51.76 (47.74–55.79)  | 0.3893  |
|                                                       | Yes                   | 287 | 48.24 (44.21–52.26)  |         |
| Down syndrome                                         | Neither Down nor Mosaic | 331 | 55.63 (51.63–59.63)  | <.0001  |
|                                                       | Down syndrome         | 254 | 42.69 (38.70–46.67)  |         |
|                                                       | Mosaic Down syndrome  | 10  | 1.68 (0.64–2.72)     |         |
| Turner syndrome                                       | Yes                   | 11  | 1.85 (0.76–2.93)     | <.0001  |
|                                                       | No                    | 584 | 98.15 (97.07–99.24)  |         |
| Child with congenital abnormalities                   | Yes                   | 11  | 1.85 (0.76–2.93)     | <.0001  |
|                                                       | No                    | 584 | 98.15 (97.07–99.24)  |         |

Table 1.
Descriptive analyses of the samples analyzed (n = 595).
| Variables                          | Categories         | Yes                      | P value | No                      | P value |
|-----------------------------------|--------------------|--------------------------|---------|-------------------------|---------|
|                                   | N                  | 95% CI                   | P       | N                       | 95% CI  | P value |
| Gender                            | Male               | 5 45.45 (10.37–80.54)    | 0.7630  | 274 46.92 (42.86–50.98) | <.0001  |
|                                   | Female             | 6 54.55 (19.46–89.63)    |         | 307 52.57 (48.51–56.63) |         |
| Religion                          | Muslim             | 2 25.00 (0.00–63.70)     | 0.1573  | 150 28.57 (24.69–32.45) | <.0001  |
|                                   | Hindu              | 6 75.00 (36.30–100.00)   |         | 375 71.43 (67.55–75.31) |         |
| History of consanguinity          | Yes                | 1 9.09 (0.00–29.35)      | 0.0067  | 27 4.62 (2.92–6.33)     | <.0001  |
|                                   | No                 | 10 90.91 (70.65–100.00)  |         | 557 95.38 (93.67–97.08) |         |
| Contraceptives used               | Yes                | 3 27.27 (0.00–58.65)     | 0.1317  | 101 17.29 (14.22–20.37) | <.0001  |
|                                   | No                 | 8 72.73 (41.35–100.00)   |         | 483 82.71 (79.63–85.78) |         |
| Addiction of father               | None               | 8 72.73 (41.35–100.00)   | 0.0201  | 333 57.02 (52.99–61.05) | <.0001  |
|                                   | Smoking            | 2 18.18 (0.00–45.36)     |         | 176 30.14 (26.40–33.87) |         |
|                                   | Smoking/Drug       | 1 9.09 (0.00–29.35)      |         | 64 10.96 (8.42–13.50)   |         |
|                                   | Smoking,Drug/Alcohol | — —                   |         | 11 1.88 (0.78–2.99)     |         |
| History of spontaneous abortion   | Yes                | 4 80.00 (24.47–100.00)   | 0.1797  | 202 86.32 (81.89–90.76) | <.0001  |
|                                   | No                 | 1 20.00 (0.00–75.53)     |         | 32 13.68 (9.24–18.11)   |         |
| Presence of diabetes              | Yes                | 1 9.09 (0.00–29.35)      | 0.0067  | 42 7.19 (5.09–9.29)     | <.0001  |
|                                   | No                 | 10 90.91 (70.65–100.00)  |         | 542 92.81 (90.71–94.91) |         |
| Presence of hormonal deficiencies | Yes                | 2 18.18 (0.00–45.36)     | 0.0348  | 60 10.27 (7.80–12.74)   | <.0001  |
| (FSH/TSH/etc.)                    | No                 | 9 81.82 (54.64–100.00)   |         | 524 89.73 (87.26–92.20) |         |
| History of congenital disease     | Yes                | 1 9.09 (0.00–29.35)      | 0.0067  | 70 11.99 (9.34–14.63)   | <.0001  |
| among first-degree relative       | No                 | 10 90.91 (70.65–100.00)  |         | 514 88.01 (85.37–90.66) |         |
| Any genetic abnormality detected  | Yes                | 7 63.64 (29.74–97.53)    | 0.3657  | 301 51.54 (47.48–55.61) | 0.4564  |
|                                   | No                 | 4 36.36 (2.47–70.26)     |         | 283 48.46 (44.39–52.52) |         |
| Down syndrome                     | Neither            | 7 63.64 (29.74–97.53)    | 0.3657  | 324 55.48 (51.44–59.52) | <.0001  |
|                                   | Mosaic             | 4 36.36 (2.47–70.26)     |         | 250 42.81 (38.78–46.83) |         |
|                                   | Down syndrome      | — —                     |         | 10 1.71 (0.66–2.77)     |         |
|                                   | Mosaic             | — —                     |         | 11 1.88 (0.78–2.99)     | <.0001  |
|                                   | Down syndrome      | — —                     |         | 573 98.12 (97.01–99.22) |         |

Table 2. Descriptive analyses regarding congenital anomalies.
3. Results

The tablewise description is as follows:

In Table 1: of the total 595 samples analyzed, 279 (46.89%) were males, 313 (52.61%) were females, and for three subjects sex could not be determined. The majority belonged to the Hindu religion (381, 71.48%) followed by Muslim (152, 28.52%). A history of consanguinity was observed among 28 (4.71%) subjects. Among females who got pregnant, 206 (86.19%) had a history of spontaneous abortion and 104 (17.48%) reported use of contraceptives, 178 (29.92%) fathers were addicted to smoking, 65 (10.92%) to both smoking and drugs, and 11 (1.85%) to either smoking or drugs or alcohol. Among total subjects, 43 (7.23%) were diagnosed with diabetes, 62 (10.42%) had some hormonal deficiencies, and 71 (11.93%) had a history of congenital disease among first-degree relatives.

More than half of the tested samples [308 (51.76%)] were from normal subjects, 254 (42.69%)

| Variables                        | Categories | Any genetic abnormality detected |
|----------------------------------|------------|----------------------------------|
|                                  |            | No                               |
|                                  |            | N   | 95% CI       | P value | N   | 95% CI       | P value |
| Gender                           | Male       | 116 | 37.66 (32.22–43.10)  | <.0001  | 163 | 56.79 (51.03–62.56) | <.0001  |
|                                  | Female     | 190 | 61.69 (56.23–67.15)  | <.0001  | 123 | 42.86 (37.10–48.62)  | <.0001  |
| Religion                         | Muslim     | 73  | 28.40 (22.85–33.96)  | <.0001  | 79  | 28.62 (23.26–33.99)  | <.0001  |
|                                  | Hindu      | 184 | 71.60 (66.04–77.15)  | <.0001  | 197 | 71.38 (66.01–76.74)  | <.0001  |
| History of consanguinity         | Yes        | 15  | 4.87 (2.45–7.29)    | <.0001  | 13  | 4.53 (2.11–6.95)  | <.0001  |
|                                  | No         | 293 | 95.13 (92.71–97.55)  | <.0001  | 274 | 95.47 (93.05–97.89)  | <.0001  |
| Contraceptives Used              | Yes        | 48  | 15.58 (11.51–19.66)  | <.0001  | 56  | 19.51 (14.90–24.12)  | <.0001  |
|                                  | No         | 260 | 84.42 (80.34–88.49)  | <.0001  | 231 | 80.49 (75.88–85.10)  | <.0001  |
| Addiction of father              | None       | 181 | 58.77 (53.24–64.29)  | <.0001  | 160 | 55.75 (49.97–61.53)  | <.0001  |
|                                  | Smoking    | 93  | 30.19 (25.04–35.35)  | <.0001  | 85  | 29.62 (24.30–34.93)  | <.0001  |
|                                  | Smoking/drug | 27  | 8.77 (5.59–11.94)  | <.0001  | 38  | 13.24 (9.30–17.19)  | <.0001  |
|                                  | Smoking/drug/alcohol | 7   | 2.27 (0.60–3.95)  | <.0001  | 4   | 1.39 (0.03–2.76)  | <.0001  |
| History of spontaneous abortion  | Yes        | 104 | 80.62 (73.71–87.53)  | <.0001  | 102 | 92.73 (87.80–97.66)  | <.0001  |
|                                  | No         | 25  | 19.38 (12.47–26.29)  | 8       | 7.27 (2.34–12.20)  | <.0001  |
| Presence of diabetes             | Yes        | 21  | 6.82 (3.99–9.65)    | <.0001  | 22  | 7.67 (4.57–10.76)  | <.0001  |
|                                  | No         | 287 | 93.18 (90.35–96.01)  | 265     | 92.33 (89.24–95.43)  | <.0001  |
| Presence of hormonal deficiencies (FSH/TSH/etc.) | Yes | 29  | 9.42 (6.14–12.70)  | <.0001  | 33  | 11.50 (7.79–15.21)  | <.0001  |
|                                  | No         | 279 | 90.58 (87.30–93.86)  | 254     | 88.50 (84.79–92.21)  | <.0001  |
| History of congenital disease among first-degree relative | Yes | 39  | 12.66 (8.93–16.40)  | <.0001  | 32  | 11.15 (7.49–14.81)  | <.0001  |
|                                  | No         | 269 | 87.34 (83.60–91.07)  | 255     | 88.85 (85.19–92.51)  | <.0001  |

Table 3. Descriptive analyses regarding congenital abnormalities.
Table 4. Descriptive analyses of samples regarding Down syndrome.

| Variables                                      | Neither Down nor Mosaic (n = 331) | Down Syndrome (n = 254) | Mosaic Down Syndrome (n = 10) |
|------------------------------------------------|----------------------------------|-------------------------|-------------------------------|
| N                                              | 95% CI                           | N 95% CI                | N 95% CI                      |
| Gender                                         | Male                             | 119 (35.95, 41.15)      | 155 (61.02, 67.08)            | 0.0004 5 (9.00, 12.30-87.70) 1.0000 |
|                                                | Female                           | 209 (63.14, 69.48)      | 243 (95.67, 93.15-98.19)      | 0.0001 5 (9.00, 12.30-87.70) 1.0000 |
| Religion                                       | Hindu                            | 136 (71.50, 75.89)      | 117 (72.24, 66.60-77.89)      | <0.0001 10 (100.00-100.00) 1.0000 |
|                                                | Muslim                           | 82 (24.50, 30.13)       | 68 (27.76, 22.13-33.40)       | <0.0001 0 (0.00-50.16) 0.0578 |
| History of consanguinity                       | Yes                              | 15 (4.53, 6.78)         | 11 (4.33, 1.81-6.85)          | <0.0001 0 (0.00-50.16) 0.0578 |
|                                                | No                               | 316 (95.47, 97.72)      | 243 (95.67, 93.15-98.19)      | <0.0001 0 (0.00-50.16) 0.0578 |
| Contraceptives used                            | Yes                              | 52 (15.71, 19.65)       | 49 (19.29, 14.43-24.19)       | <0.0001 0 (0.00-50.16) 0.0578 |
|                                                | No                               | 279 (84.29, 80.35-88.23) | 206 (80.71, 75.82-85.59)     | <0.0001 0 (0.00-50.16) 0.0578 |
| Smoking                                        | Yes                              | 101 (42.12-68.79)       | <0.0001 11 (13.13-48.19)      | <0.0001 0 (0.00-50.16) 0.0578 |
|                                                | No                               | 316 (95.47, 97.72)      | 243 (95.67, 93.15-98.19)      | <0.0001 0 (0.00-50.16) 0.0578 |
| History of spontaneous abortion                | Yes                              | 107 (31.50, 35.35-37.50) | 2 (3.06, 0.36-6.45)           | <0.0001 0 (0.00-50.16) 0.0578 |
|                                                | No                               | 264 (68.50, 64.65-72.38) | 2 (3.06, 0.36-6.45)           | <0.0001 0 (0.00-50.16) 0.0578 |
| Presence of diabetes                           | Yes                              | 26 (7.96, 12.12-17.26)  | 22 (8.66, 5.05-13.34)         | <0.0001 0 (0.00-50.16) 0.0578 |
|                                                | No                               | 310 (92.04, 97.33-94.05) | 25 (22.5, 84.65-92.52)        | <0.0001 0 (0.00-50.16) 0.0578 |
| Presence of hormonal deficiencies (FSH/TSH etc.) | Yes                             | 30 (9.06, 5.85-13.59)   | 20 (10.00, 0.00-100.00)       | <0.0001 0 (0.00-50.16) 0.0578 |
|                                                | No                               | 301 (90.94, 94.19-97.56) | 205 (90.00, 84.65-92.52)      | <0.0001 0 (0.00-50.16) 0.0578 |

6
were identified as Down syndrome, 10 (1.68%) as mosaic Down syndrome, while 11 (1.85%) as Turner syndrome, and 11 (1.85%) children with other congenital anomalies.

In Table 2: of the total 11 children with congenital abnormalities, five (45.45%) were males. Based on the available information, it was observed that six (75%) belonged to the Hindu religion followed by Muslim (2, 28.52%), one (9.09%) had a history of consanguinity, four (80%) had a history of spontaneous abortion, three (27.27%) reported use of contraceptives, two fathers (18.18%) were addicted to smoking, one (9.09%) was addicted to both smoking and drugs, one subject (9.09%) was diagnosed with diabetes, two subjects (18.18%) with hormonal deficiencies, one subject (9.09%) had a history of congenital disease among first-degree relatives, four (36.36%) were identified as Down syndrome, and none of them with Turner syndrome.

| Variables                          | Categories | Diagnosed with Turner syndrome |  |
|------------------------------------|------------|---------------------------------|---|
|                                   | Yes | N     | 95% CI | P value | No | N     | 95% CI | P value |
| Gender                            | Male — | — | — | — | 279 | 47.77 (43.71–51.84) | <.0001 |
|                                   | Female 11 | 100.00 (100.00–100.00) | 302 | 51.71 (47.65–55.78) |  |
| Religion                          | Muslim 4 | 40.00 (3.06–76.94) | 0.5271 | 148 | 28.30 (24.43–32.17) | <.0001 |
|                                   | Hindu 6 | 60.00 (23.06–96.94) | 375 | 71.70 (67.83–75.57) |  |
| History of consanguinity          | Yes — | — | — | — | 28 | 4.79 (3.06–6.53) | <.0001 |
|                                   | No 11 | 100.00 (100.00–100.00) | 556 | 95.21 (93.47–96.94) |  |
| Contraceptive used                | Yes 2 | 18.18 (0.00–45.36) | 0.0348 | 102 | 17.47 (14.38–20.55) | <.0001 |
|                                   | No 9 | 81.82 (54.64–100.00) | 482 | 82.53 (79.45–85.62) |  |
| Addiction of father               | None 5 | 45.45 (10.37–80.54) | 0.5292 | 336 | 57.53 (53.51–61.55) | <.0001 |
|                                   | Smoking 4 | 36.36 (2.47–70.26) | 174 | 29.79 (26.07–33.51) |  |
|                                   | Smoking/ drug 2 | 18.18 (0.00–45.36) | 63 | 0.79 (8.26–13.31) |  |
|                                   | Smoking/ drug/ alcohol — — — 11 | 1.88 (0.78–2.99) |  |
| History of spontaneous abortion   | Yes 1 | 50.00 (0.00–100.00) | 1.0000 | 205 | 86.50 (82.12–90.88) | <.0001 |
|                                   | No 1 | 50.00 (0.00–100.00) | 32 | 13.50 (9.12–17.88) |  |
| Presence of diabetes              | Yes — | — | — | — | 43 | 7.36 (5.24–9.49) | <.0001 |
|                                   | No 11 | 100.00 (100.00–100.00) | 541 | 92.64 (90.51–94.76) |  |
| Presence of hormonal deficiencies (FSH/TSH/ etc.) | Yes — | — | — | — | 62 | 10.62 (8.31–13.12) | <.0001 |
|                                   | No 11 | 100.00 (100.00–100.00) | 522 | 89.38 (86.88–91.89) |  |
| History of congenital disease among first degree relative | Yes 1 | 9.09 (0.00–29.35) | 0.0067 | 70 | 11.99 (9.34–14.63) | <.0001 |
|                                   | No 10 | 90.91 (70.65–100.00) | 514 | 88.01 (85.37–90.66) |  |

Table 5. Descriptive analyses of participants regarding turner syndrome (n = 11).
In Table 3: of the 283 samples tested to have some genetic abnormalities, 163 (56.79%) were males, 197 (71.38%) belonged to the Hindu religion followed by Muslim (79, 28.62%), 13 (4.53%) had a history of consanguinity, 102 (92.73%) had a history of spontaneous abortion, 56 (19.51%) reported use of contraceptives, 85 fathers (29.62%) were addicted to smoking, 38 (13.24%) to both smoking and drugs, and 4 (1.39%) to either smoking or drugs or alcohol. Among these 283 subjects, 22 (7.67%) were diagnosed with diabetes, 33 (11.50%) had some hormonal deficiencies, and 32 (11.15%) had a history of congenital disease among first-degree relatives.

In Table 4: Among the total 254 samples who were diagnosed with Down syndrome, 155 (61.02%) were males, 177 (72.24%) belonged to the Hindu religion followed by Muslim (68, 27.76%), 11 (4.33%) had a history of consanguinity, 94 (93.07%) had a history of spontaneous abortion, 49 (19.29%) couples reported use of contraceptives, 85 fathers (29.62%) were addicted to smoking, 38 (13.24%) to both smoking and drugs, and 4 (1.39%) to either smoking or drugs or alcohol. Among these 283 subjects, 22 (7.67%) were diagnosed with diabetes, 33 (11.50%) had some hormonal deficiencies, and 32 (11.15%) had a history of congenital disease among first-degree relatives.

In Table 5: among the total 11 samples who were diagnosed with Turner syndrome and all of them were females, six (60.00%) belonged to the Hindu religion.
followed by Muslim (4, 40.00%) and none had a history of consanguinity. One (50.00%) had a history of spontaneous abortion, two (18.18%) couples reported use of contraceptives, four (36.36%) fathers were addicted to smoking, two (18.18%) to both smoking and drugs, none were diagnosed with diabetes or hormonal deficiencies, and one (9.09%) had a history of congenital disease among first-degree relatives.

In Table 6: compared to females, males were 54% (odds ratio, OR = 0.46, 95% CI = 0.33–0.64) less likely to be normal. Additionally, for females who got pregnant and had a history of spontaneous abortion, the chance of being normal was 67% less (odds ratio, OR = 0.33, 95% CI = 0.14–0.76) compared to those who did not have such history.

In Table 7: compared to females, males were almost thrice likely (odds ratio, OR = 2.75, 95% CI = 1.96–3.86) to be clinically diagnosed with Down syndrome. Additionally, in females who got pregnant and had a history of spontaneous abortion, the risk of Down syndrome was more than three times higher (odds ratio, OR = 3.26, 95% CI = 1.35–7.86) than those who did not have such history. Subjects with a history of consanguinity had a four times higher risk of being clinically diagnosed with mosaic Down syndrome (odds ratio, OR = 4.29, 95% CI = 1.06–17.50) than those who have no such history. Additionally, history of smoking and drug addiction among fathers was positively (odds ratio, OR = 0.49, 95% CI = 0.13–1.87) associated with a higher likelihood of mosaic Down syndrome than those who did not have such history. Moreover, the risk of being diagnosed with this
A defect was fourfold (odds ratio, OR = 4.30, 95% CI = 1.06–17.50) among participants detected with some hormonal deficiencies than those who did not have such deficiencies.

### Table 8. Predictors of Turner syndrome.

| Variables                                              | Categories | Diagnosed with Turner syndrome (ref = no) |
|--------------------------------------------------------|------------|------------------------------------------|
|                                                        |            | Yes                                      |
| Gender (ref = female)                                  | Male       | —                                        |
| Religion (ref = Muslim)                                | Hindu      | 0.59 (0.17–2.13) 0.4219                  |
| History of consanguinity (ref = no)                    | Yes        | —                                        |
| Contraceptive used (ref = no)                          | Yes        | 1.05 (0.22–4.93) 0.9506                  |
| Addiction of father (ref = none)                       | Smoking    | 1.55 (0.41–5.83) 0.5208                  |
|                                                        | Smoking/drug | 2.13 (0.41–11.24) 0.3715                |
|                                                        | Smoking/drug/alcohol | —  —                                  |
| History of spontaneous abortion (ref = no)             | Yes        | 0.16 (0.01–2.56) 0.1930                  |
| Presence of diabetes (ref = no)                        | Yes        | —                                        |
| Presence of hormonal deficiencies (FSH/TSH/etc.) (ref = no) | Yes        | —                                        |
| History of congenital disease among first degree relative (ref = no) | Yes | 0.74 (0.09–5.82) 0.7704                  |

### Table 9. Predictors having congenital abnormalities.

| Variables                                              | Categories | Child with congenital abnormalities (ref = no) |
|--------------------------------------------------------|------------|-----------------------------------------------|
|                                                        |            | Yes                                          |
| Gender (ref = female)                                  | Male       | 0.93 (0.28–3.09) 0.9106                      |
| Religion (ref = Muslim)                                | Hindu      | 1.20 (0.24–6.01) 0.8245                      |
| History of consanguinity (ref = no)                    | Yes        | 2.06 (0.26–16.71) 0.4971                     |
| Contraceptives used (ref = no)                         | Yes        | 1.79 (0.47–6.88) 0.3944                      |
| Addiction of father (ref = none)                       | Smoking    | 0.47 (0.10–2.25) 0.3470                      |
|                                                        | Smoking/drug | 0.65 (0.08–5.29) 0.6875                    |
|                                                        | Smoking/drug/alcohol | —  —                                  |
| History of spontaneous abortion (ref = no)             | Yes        | 0.63 (0.07–5.85) 0.6875                      |
| Presence of diabetes (ref = no)                        | Yes        | 1.29 (0.16–10.32) 0.8097                     |
| Presence of hormonal deficiencies (FSH/TSH/etc.) (ref = no) | Yes        | 1.94 (0.41–9.19) 0.4033                      |
| History of congenital disease among first degree relative (ref = no) | Yes | 0.74 (0.09–5.82) 0.7704                      |
In Table 8: although all the predictors such as male gender, Hindu religion, positive history of consanguinity, history of having the spontaneous abortion, contraceptives use, addiction of father, the presence of diabetes or some hormonal deficiencies and having a history of congenital disease among first-degree relatives seemed to be positively associated with the risk of Turner syndrome, results were not statistically significant due to small sample size and lack of power.

In Table 9: the other congenital anomalies did not show any association with the studied factors and results were not statistically significant due to the small sample size and lack of power. Thus, for inconclusive and empirical evidence regarding predictors of participants having a child with congenital abnormalities, a large sample size is required.

4. Discussion

In this study, the different factors such as gender, age, ethnicity, addiction, hormonal status have been analyzed to investigate their possible effect on Down syndrome, Turner syndrome, and other congenital disease prevalence. The distributions of the sample characteristics were significantly different across strata of gender, religion, history of consanguinity, contraceptive used, the addiction of participants’ father, whether diagnosed with diabetes or hormonal deficiencies or Down syndrome or Turner syndrome, and history of congenital disease among first-degree relatives and child with congenital abnormalities. The distributions of the children with congenital abnormalities such as ambiguous genitalia or syndactyly were significantly different across strata of history of consanguinity, addiction of parent, whether diagnosed with diabetes or hormonal deficiencies or Down syndrome or Turner syndrome and history of congenital disease among first-degree relatives. Distributions of sample characteristics were significantly different across strata of gender, religion, history of consanguinity, contraceptive used, history of spontaneous abortion, addiction of father, whether diagnosed with diabetes or hormonal deficiencies, and history of congenital disease among first-degree relatives (Table 3). The distributions of sample characteristics who were clinically diagnosed with Down syndrome were significantly different across strata of gender, religion, history of consanguinity, contraceptive used, addiction of father, whether diagnosed with diabetes or hormonal deficiencies, and history of congenital disease among first-degree relatives whether individuals diagnosed with mosaic Down syndrome were not significantly different across strata of those factors. Except for the use of contraceptives, distributions of the sample characteristics who were clinically diagnosed with Turner syndrome were not significantly different across the strata of gender, religion, history of consanguinity, addiction of father, whether diagnosed with diabetes or hormonal deficiencies, and history of congenital disease among first-degree relatives. Other predictors, such as Hindu religion, positive history of consanguinity, use of contraceptives, addiction of father, presence of diabetes or hormonal deficiencies, and having a history of congenital disease among first-degree relatives, seemed more likely to be clinically diagnosed as normal but results were not statistically significant due to small sample size and lack of power. Thus, for inconclusive and empirical evidence regarding predictors of clinically normal subjects, a large sample size is required.

On the basis of outcomes, the possible effects of sociodemographic factors are convenient regarding the studied congenital disease occurrence, though a large-scale analysis from all aspects is needed.
5. Conclusion

In this chapter, we have found that some factors such as age, addictions, hormonal imbalances are likely to be associated with Down syndrome, Turner syndrome, and also the other studied congenital diseases. There are several sociodemographic factors that seem to be associated with these congenital disorders, though a large sample size is required for better assessment.

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