Chapter

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 mamgement 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			No		
		N	95% CI	P value	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 (FSH/TSH/ etc.)	Yes	2	18.18 (0.00–45.36)	0.0348	60	10.27 (7.80–12.74)	<.0001
	No	9	81.82 (54.64–100.00)		524	89.73 (87.26–92.20)	
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)	
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 down nor Mosaic	7	63.64 (29.74–97.53)	0.3657	324	55.48 (51.44–59.52)	<.0001
	Down syndrome	4	36.36 (2.47–70.26)		250	42.81 (38.78–46.83)	
	Mosaic Down syndrome	—	—	—	10	1.71 (0.66–2.77)	
Turner syndrome	Yes	—	—	—	11	1.88 (0.78–2.99)	<.0001
	No	11	100.00 (100.00–100.00)	—	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			Yes		
		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)		123	42.86 (37.10–48.62)	
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)		197	71.38 (66.01–76.74)	
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)		274	95.47 (93.05–97.89)	
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)		231	80.49 (75.88–85.10)	
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)		85	29.62 (24.30–34.93)	
	Smoking/ drug	27	8.77 (5.59–11.94)		38	13.24 (9.30–17.19)	
	Smoking/ drug/ alcohol	7	2.27 (0.60–3.95)		4	1.39 (0.03–2.76)	
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)	
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)	
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)	
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)	

Table 3.
Descriptive analyses regarding congenital abnormalities.

Variables	Categories	Diagnosed with								
		Neither Down nor Mosaic (n = 331)			Down syndrome (n = 254)			Mosaic Down syndrome (n = 10)		
		N	95% CI	p value	N	95% CI	P value	N	95% CI	P value
Gender	Male	119	35.95 (30.76–41.15)	<.0001	155	61.02 (54.99–67.06)	0.0004	5	50.00 (12.30–87.70)	1.0000
	Female	209	63.14 (57.92–68.37)		99	38.98 (32.94–45.01)		5	50.00 (12.30–87.70)	
Religion	Muslim	82	29.50 (24.10–34.89)	<.0001	68	27.76 (22.11–33.40)	<.0001	2	20.00 (0.00–50.16)	0.0578
	Hindu	196	70.50 (65.11–75.90)		177	72.24 (66.60–77.89)		8	80.00 (49.84–100.00)	
History of consanguinity	Yes	15	4.53 (2.28–6.78)	<.0001	11	4.33 (1.81–6.85)	<.0001	2	20.00 (0.00–50.16)	0.0578
	No	316	95.47 (93.22–97.72)		243	95.67 (93.15–98.19)		8	80.00 (49.84–100.00)	
Contraceptives used	Yes	52	15.71 (11.77–19.65)	<.0001	49	19.29 (14.41–24.18)	<.0001	3	30.00 (0.00–64.56)	0.2059
	No	279	84.29 (80.35–88.23)		205	80.71 (75.82–85.59)		7	70.00 (35.45–100.00)	
Addiction of father	None	191	57.70 (52.35–63.05)	<.0001	147	57.87 (51.76–63.99)	<.0001	3	30.00 (0.00–64.56)	0.9048
	Smoking	101	30.51 (25.53–35.50)		73	28.74 (23.14–34.34)		4	40.00 (3.06–76.94)	
	Smoking/drug	31	9.37 (6.21–12.52)		31	12.20 (8.15–16.26)		3	30.00 (0.00–64.56)	
	Smoking/drug/alcohol	8	2.42 (0.75–4.08)		3	1.18 (0.00–2.52)		—	—	—
History of spontaneous abortion	Yes	107	80.45 (73.62–87.28)	<.0001	94	93.07 (88.03–98.11)	<.0001	5	100.00 (100.00–100.00)	—
	No	26	19.55 (12.72–26.38)		7	6.93 (1.89–11.97)		—	—	
Presence of diabetes	Yes	21	6.34 (3.70–8.98)	<.0001	22	8.66 (5.18–12.14)	<.0001	—	—	—
	No	310	93.66 (91.02–96.30)		232	91.34 (87.86–94.82)		10	100.00 (100.00–100.00)	
Presence of hormonal deficiencies (FSH/TSH/etc.)	Yes	30	9.06 (5.95–12.17)	<.0001	29	11.42 (7.48–15.35)	<.0001	3	30.00 (0.00–64.56)	0.2059
	No	301	90.94 (87.83–94.05)		225	88.58 (84.65–92.52)		7	70.00 (35.45–100.00)	
History of congenital disease among first degree relative	Yes	40	12.08 (8.55–15.61)	<.0001	29	11.42 (7.48–15.35)	<.0001	2	20.00 (0.00–50.16)	0.0578
	No	291	87.92 (84.39–91.45)		225	88.58 (84.65–92.52)		8	80.00 (49.84–100.00)	

Table 4.
Descriptive analyses of samples regarding down syndrome.

Variables	Categories	Diagnosed with Turner syndrome					
		Yes			No		
		N	95% CI	P value	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 (8.11–13.12)	<.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).

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 as normal (ref = no)	
		Yes	
		OR (95% CI)	P value
Gender (ref = female)	Male	**0.46 (0.33–0.64)**	**<.0001**
Religion (ref = Muslim)	Hindu	1.01 (0.69–1.47)	0.9555
History of consanguinity (ref = no)	Yes	1.08 (0.50–2.31)	0.8449
History of spontaneous abortion (ref = no)	Yes	**0.33 (0.14–0.76)**	**0.0091**
Contraceptives used (ref = no)	Yes	0.76 (0.50–1.16)	0.2084
Addiction of father (ref = none)	Smoking	0.97 (0.67–1.39)	0.8570
	Smoking/drug	0.63 (0.37–1.08)	0.0897
	Smoking/drug/alcohol	1.55 (0.45–5.38)	0.4928
Presence of diabetes (ref = no)	Yes	0.88 (0.47–1.64)	0.6899
Presence of hormonal deficiencies (FSH/TSH/etc) (ref = no)	Yes	0.80 (0.47–1.36)	0.4067
History of congenital disease among first degree relative (ref = no)	Yes	1.16 (0.70–1.90)	0.5710

Table 6.
Predictors of participants who were diagnosed as normal.

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, 73 (28.74%) fathers were addicted to smoking, 31 (12.20%) to both smoking and drugs and 3 (1.18%) to either smoking or drugs or alcohol, 22 (8.66%) were diagnosed with diabetes, 29 (11.42%) had some hormonal deficiencies, and 29 (11.42%) had a history of congenital disease among first-degree relatives. Among 10 samples who were diagnosed with mosaic Down syndrome, five (50.00%) were males, eight (80%) belonged to the Hindu religion followed by Muslim (2, 20.00%), and two (20.00%) had a history of consanguinity, all had a history of spontaneous abortion, three (30.00%) reported use of contraceptives, four (40.00%) were addicted to smoking, three (30.00%) to both smoking and drugs, while none of them were diagnosed with diabetes, three (30.00%) had some hormonal deficiencies, and two (20.00%) 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

Variables	Categories	Clinical diagnosed with (ref = neither Down or Mosaic)			
		Down syndrome		Mosaic down syndrome	
		OR (95% CI)	P value	OR (95% CI)	P value
Gender (ref = female)	Male	**2.75 (1.96–3.86)**	**<.0001**	1.76 (0.50–6.19)	0.3809
Religion (ref = Muslim)	Hindu	1.09 (0.74–1.59)	0.6604	1.67 (0.35–8.05)	0.5206
History of consanguinity (ref = no)	Yes	0.95 (0.43–2.11)	0.9069	**5.27 (1.03–26.98)**	**0.0462**
Contraceptives used (ref = no)	Yes	1.28 (0.83–1.97)	0.2567	2.30 (0.58–9.18)	0.2384
Addiction of father (ref = none)	Smoking	0.94 (0.65–1.36)	0.7393	2.52 (0.55–11.49)	0.2319
	Smoking/ drug	1.30 (0.76–2.24)	0.3440	**6.16 (1.19–31.91)**	**0.0302**
	Smoking/ drug/ alcohol	0.49 (0.13–1.87)	0.2945	—	—
History of spontaneous abortion (ref = no)	Yes	**3.26 (1.35–7.86)**	**0.0084**	—	—
Presence of diabetes (ref = no)	Yes	1.40 (0.75–2.61)	0.2890	—	—
Presence of hormonal deficiencies (FSH/TSH/ etc) (ref = no)	Yes	1.29 (0.75–2.22)	0.3497	**4.30 (1.06–17.50)**	**0.0416**
History of congenital disease among first degree relative (ref = no)	Yes	0.94 (0.56–1.56)	0.8042	1.82 (0.37–8.87)	0.4593

Table 7.
Predictors of down syndrome.

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 = 0.33, 95% CI = 0.14–0.76) 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 = 5.27, 95% CI = 1.03–26.98) than those who have no such history. Additionally, history of smoking and drug addiction among fathers was positively (odds ratio, OR = 6.16, 95% CI = 1.19–31.91) 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

Variables	Categories	Diagnosed with Turner syndrome (ref = no)	
		Yes	
		OR (95% CI)	P value
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 8.
Predictors of turner syndrome.

Variables	Categories	Child with congenital abnormalities (ref = no)	
		Yes	
		OR (95% CI)	P value
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

Table 9.
Predictors having congenital abnormalities.

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.

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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