Announcements

The 16th National Birth Defects Prevention Day — September 12, 2020

Birth defects (BDs) can be defined as abnormalities of structure or function that occur during intrauterine life and can be identified prenatally, after birth, or later in infancy (*1*). The World Health Organization (WHO) indicated that every year an estimated 6% of babies worldwide are born with a BD, and over 300,000 deaths occur in infants within 4 weeks due to BDs (*2*). BDs are a global problem, and China has a high incidence of BDs as every year an estimated 0.9 million children — 6% of total national births — are born with BDs (*3*). BDs can contribute to long-term disability, which may have significant impacts on individuals, families, healthcare systems, and societies.

Fortunately, experience showed that about 70% of BDs can either be prevented or that affected children can be offered care (*4*). Therefore, in 2005, the Chinese government declared that September 12 would annually be "National Birth Defects Prevention Day" (NBDPD) to raise awareness of this serious problem and advocate for more BDs prevention, surveillance, care, and research. This year, NBDPD has become even more significant because of the coronavirus disease 2019 (COVID-19) pandemic. Therefore, the theme of the coming 16th NBDPD in 2020 will be "United against the pandemic, safeguarding new life."

doi: 10.46234/ccdcw2020.194

Submitted: September 03, 2020; Accepted: September 04, 2020

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Vital Surveillances

National Perinatal Prevalence of Selected Major Birth Defects — China, 2010–2018

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ABSTRACT

Introduction: An estimated of 900,000 infants are born with birth defects each year in China causing a substantial disease burden. This study aimed to depict the epidemiological patterns of selected major birth defects in Chinese perinatal births and provide important baseline data for future prevention.

Methods: Data from the Chinese Birth Defects Monitoring Network (CBDMN) during 2010–2018 were used to analyze the epidemiological pattern in the prevalence of 15 major birth defects and the trends over time.

Results: In the period of 2010–2018, the top 10 most frequently-occurring birth defects in China included congenital heart diseases (CHDs), polydactyly, cleft lip with or without palate (CL/P), club foot, syndactyly, hydrocephalus, hypospadias, limb reduction defects (LRD), anotia/microtia, and anorectal atresia/stenosis. There was a decrease in the prevalence of neural tube defects, CL/P, hydrocephalus, LRD, gastroschisis, and omphalocele, but there were increases in the prevalence of CHDs, cleft palate, hypospadias, club foot, polydactyly, and syndactyly. The prevalence of most birth defects varied significantly by maternal age, area types (urban/rural), and geographic regions.

Conclusions and Implications for Public Health Practice: The findings indicated that the comprehensive prevention of birth defects should focus on these selected birth defects, elderly pregnant women, rural areas, and western regions.

INTRODUCTION

Birth defects (BDs) affect 4%–8% of births worldwide (*1*). The estimated prevalence in China is

5.6%, with approximately 900,000 infants born with various defects each year (*2*). BDs are the leading cause of infant mortality in China, accounting for about 20% of deaths (*2–3*). Other adverse outcomes include premature death in early life, disabilities, impaired physical and mental well-being, negative effects on quality of life, which result in a substantial disease burden on families and societies (*1–2*,*4*). According to the Global Burden of Disease Study in 2017, BDs are the tenth leading cause of disability-adjusted life years (DALYs) among women and ninth among men (*5*). As a global public health problem, BDs have been of major concern to governments, health professionals, and the public.

Although several genetic, behavioral, and environmental risk factors have been identified, the underlying causes of most BDs were unclear (*1–2*). BDs surveillance played an important role in exploring possible risks, monitoring dynamic changes in prevalence, and providing evidence for interventions (*4*). With rapid socioeconomic development in China, birth policies, family income, maternal nutrition, prenatal care, and socioeconomic and demographic factors have varied significantly. However, reliable data on the prevalence of major BDs was limited. Using 2010–2018 data extracted from the Chinese Birth Defects Monitoring Network (CBDMN) (*4*,*6–8*), we aimed to analyze the epidemiological pattern in the prevalence of 15 major BDs and their trends over time.

METHODS

CBDMN is a nationwide hospital-based BDs surveillance network that covers 763 member hospitals in 327 counties or districts in 31 provincial-level administrative divisions (PLADs). CDBMN collects information on live births, stillbirths, and elective terminations of pregnancies at or above 28 weeks of gestation occurring in member hospitals. More than 2 million births were covered each year representing over 10% of live births in China (*6*). All anomalies diagnosed during the perinatal period (from 28 weeks of gestation to Day 7 after birth) are required to be included in the system and coded by health professionals according to the Tenth Revision of International Classification of Diseases (ICD-10). Details of data collection and quality control have been described previously (*4*,*6–8*).

We selected 15 frequently-occurring major BDs in China for the current analysis, including neural tube defects (NTDs, Q00, Q01, and Q05), hydrocephalus (Q03), anotia/microtia (Q16.0 and Q17.2), congenital heart diseases (CHDs, Q20–Q26), cleft palate (CP, Q35), cleft lip with or without palate (CL/P, Q36–Q37), anorectal atresia/stenosis (Q42), hypospadias (Q54), club foot (Q66.0), polydactyly (Q69), syndactyly (Q70), limb reduction defects (LRD, Q71–Q73), omphalocele (Q79.2), gastroschisis (Q79.3), and Down syndrome (Q90). The perinatal prevalence rate was defined as the number of cases per 10,000 live and still perinatal births in the specified period. We calculated the prevalence rates by calendar year, maternal age (<20, 20–24, 25–29, 30–34, and ≥ 35 years), infant sex (female *vs.* male), maternal residence area type (urban/rural), and geographic regions (eastern, central, and western). The rules for urban/rural and geographic classifications in the CBDMN were described previously (*6–8*).

We used R 3.5.3 (R Development Core Team 2019) for data cleaning and analysis. Pearson chi-squared tests were used to examine differences of prevalence between various groups, and linear chi-squared tests were used to determine the time trends. The 95% confidence intervals (95% CI) for prevalence rates were estimated according to Poisson distribution. The statistical significance level (α) was set at 0.05.

RESULTS

From 2010 to 2018, a total of 18,040,393 perinatal births were recorded, of whom 52.8% were males and 47.1% were females. Overall, 2.2% of births were born to women <20 years, and 11.3% were born to women ≥35 years. The newborns whose mothers resided in rural areas accounted for 45.7%, while infants whose mothers lived in urban areas accounted for 54.3%; 32.1% of births were born to women residents of the eastern region, while 37.2% and 30.7% of births were born to women in the central and western regions, respectively.

As shown in Table 1 and Table 2, the top 10 most frequently occurring BDs included CHDs, polydactyly, CL/P, club foot, syndactyly, hydrocephalus, hypospadias, LRD, anotia/microtia, and anorectal atresia/stenosis. The prevalence rates of selected major BDs varied significantly by maternal age, maternal residence, infant sex, and geographic regions. We found a higher prevalence rate in the advanced maternal age group for CHDs, CP, hypospadias, and Down syndrome, whereas gastroschisis prevalence in the younger maternal-age group (<20 years of old) was much higher. Regarding

TABLE 1. Prevalence* (95% CI) of selected major birth defects by maternal age and infant sex in China, 2010–2018.

Item	Maternal age at delivery (years)					Infant sex		Total
	<20	20–24	25–29	30–34	≥35	Female	Male	
NTDs	8.9(8.0–9.8)	3.9(3.8–4.2)	2.0(1.9–2.1)	2.1(2.0–2.3)	3.1(2.9–3.4)	3.1(3.0–3.2)	2.3(2.3–2.5)	2.7(2.7–2.8)
Anencephalus	2.9(2.4–3.4)	1.0(0.9–1.1)	0.5(0.4–0.5)	0.5(0.4–0.6)	0.8(0.7–0.9)	0.8(0.7–0.9)	0.5(0.5–0.6)	0.7(0.6–0.7)
Spina Bifida	4.8(4.2–5.6)	2.4(2.3–2.6)	1.3(1.2–1.4)	1.3(1.2–1.4)	1.9(1.7–2.1)	1.8(1.8–1.9)	1.5(1.4–1.6)	1.7(1.6–1.7)
Encephalocele	1.2(0.9–1.6)	0.6(0.5–0.7)	0.3(0.3–0.3)	0.3(0.3–0.4)	0.4(0.3–0.5)	0.4(0.4–0.5)	0.3(0.3–0.4)	0.4(0.4–0.4)
Hydrocephalus	8.7(7.8–9.6)	6.2(5.9–6.4)	4.6(4.4–4.7)	4.7(4.5–4.9)	5.4(5.1–5.8)	4.6(4.5–4.8)	5.5(5.4–5.7)	5.1(5.0–5.2)
Anotia/microtia	2.9(2.4–3.5)	2.7(2.6–2.9)	2.7(2.6–2.9)	3.0(2.8–3.1)	3.3(3.0–3.5)	2.4(2.3–2.5)	3.3(3.2–3.4)	2.9(2.8–2.9)
CHDs	45.8(43.8–48.0)	47.7(47.0–48.4)	57.7(57.2–58.2)	64.4(63.6–65.1)	76.4(75.2–77.6)	57.8(57.3–58.3)	60.3(59.8–60.8)	59.2(58.9–59.6)
TGA	0.7(0.4–1.0)	0.7(0.6–0.8)	0.8(0.7–0.8)	0.8(0.8–0.9)	1.0(0.9–1.2)	0.6(0.5–0.6)	1.0(1.0–1.1)	0.8(0.8–0.9)
VSD	8.0(7.2–8.9)	9.2(8.9–9.5)	11.3(11.1–11.5)	12.7(12.3–13.0)	15.9(15.3–16.4)	12.3(12.1–12.5)	11.1(10.9–11.3)	11.7(11.5–11.8)
ASD	29.3(27.7–31.0)	28.6(28.1–29.2)	36.3(35.9–36.8)	41.0(40.5–41.7)	48.7(47.8–49.7)	36.1(35.7–36.5)	38.2(37.8–38.6)	37.2(36.9–37.5)
AVSD	1.8(1.4–2.3)	1.6(1.5–1.7)	1.6(1.5–1.7)	1.7(1.6–1.8)	2.2(2.0–2.4)	1.8(1.7–1.9)	1.6(1.5–1.7)	1.7(1.6–1.8)
TOF	0.9(0.6–1.3)	1.3(1.2–1.4)	1.2(1.1–1.3)	1.4(1.3–1.5)	2.0(1.8–2.2)	1.2(1.2–1.3)	1.4(1.4–1.5)	1.3(1.3–1.4)
PDA	15.3(14.2–16.6)	15.7(15.3–16.1)	20.1(19.8–20.4)	22.4(22.0–22.8)	26.8(26.1–27.5)	19.7(19.4–20.0)	21.1(20.8–21.4)	20.4(20.2–20.6)
CP	2.3(1.8–2.8)	2.4(2.3–2.6)	2.6(2.5–2.8)	2.7(2.6–2.9)	2.9(2.7–3.2)	3.3(3.2–3.4)	2.1(2.0–2.2)	2.6(2.6–2.7)
CL/P	16.5(15.3–17.8)	11.0(10.7–11.4)	6.8(6.6–7.0)	7.0(6.7–7.2)	9.3(8.9–9.7)	6.8(6.6–7.0)	9.4(9.2–9.6)	8.2(8.1–8.4)
CL	6.0(5.2–6.8)	4.3(4.1–4.6)	2.9(2.8–3.0)	3.0(2.8–3.1)	3.6(3.4–3.9)	2.8(2.7–3.0)	3.8(3.7–3.9)	3.3(3.3–3.4)
CLP	10.6(9.6–11.6)	6.7(6.4–6.9)	4.0(3.8–4.1)	4.0(3.8–4.2)	5.6(5.3–6.0)	3.9(3.8–4.1)	5.7(5.5–5.8)	4.9(4.8–5.0)
Anorectal atresia/stenosis	3.2(2.7–3.8)	2.9(2.7–3.0)	2.5(2.4–2.7)	2.8(2.7–3.0)	3.7(3.4–4.0)	1.9(1.8–2.0)	3.5(3.4–3.7)	2.8(2.8–2.9)
Hypospadias§	3.8(3.2–4.4)	4.0(3.9–4.3)	4.9(4.7–5.0)	5.6(5.4–5.9)	6.7(6.3–7.0)	–	9.6(9.4–9.8)	5.1(5.0–5.2)
Club foot	9.4(8.4–10.4)	6.5(6.2–6.8)	5.7(5.5–5.8)	5.5(5.3–5.7)	6.3(6.0–6.7)	5.7(5.6–5.9)	6.1(6.0–6.3)	5.9(5.8–6.1)
Polydactyly	23.0(21.6–24.5)	17.7(17.3–18.1)	17.1(16.8–17.4)	17.0(16.6–17.4)	18.6(18.0–19.2)	14.2(13.9–14.4)	20.4(20.1–20.7)	17.5(17.3–17.7)
Syndactyly	5.9(5.1–6.7)	5.2(4.9–5.4)	5.6(5.5–5.8)	6.0(5.8–6.3)	5.7(5.4–6.1)	4.9(4.7–5.0)	6.3(6.2–6.5)	5.7(5.5–5.8)
LRD	4.8(4.1–5.5)	3.6(3.5–3.9)	3.0(2.9–3.1)	3.1(3.0–3.3)	4.0(3.7–4.2)	2.9(2.8–3.0)	3.5(3.4–3.7)	3.3(3.2–3.4)
Omphalocele†	1.5(1.1–1.9)	0.9(0.8–1.0)	0.8(0.8–0.9)	1.0(0.9–1.1)	1.5(1.4–1.7)	1.0(0.9–1.0)	0.9(0.9–1.0)	1.0(0.9–1.0)
Gastroschisis†	5.3(4.6–6.1)	1.6(1.5–1.7)	0.5(0.4–0.5)	0.4(0.3–0.5)	0.6(0.5–0.7)	0.7(0.7–0.8)	0.8(0.7–0.9)	0.8(0.8–0.8)
Down syndrome	0.6(0.4–0.9)	0.8(0.7–0.9)	1.0(0.9–1.1)	1.3(1.2–1.5)	4.7(4.4–5.0)	1.3(1.2–1.4)	1.6(1.5–1.7)	1.5(1.4–1.5)

Abbreviations: CI=confidence interval; NTDs=neural tube defects; CHDs=congenital heart diseases; TGA=transposition of great arteries; VSD=ventricular septal defect; ASD=atrial septal defect; AVSD=atrioventricular septal defect; TOF=tetralogy of Fallot; PDA=patent ductus arteriosus; CP=cleft palate; CL/P=cleft lip with or without palate; CL=cleft lip without palate; CLP=cleft lip with palate; LRD=limb reduction defects.

* Per 10,000 perinatal births.

† Except omphalocele and gastroschisis, all selected major birth defects varied significantly by infant sex (p<0.01); All selected major birth defects varied significantly by maternal age (p<0.01).

§ The prevalence rate of hypospadias was defined as the number of case per 10,000 male perinatal births during the specified period.

TABLE 2. Prevalence* (95% CI) of selected major birth defects by urban-rural areas and geographic regions in China, 2010–2018.

Item	Maternal residence		Geographic location			Total
	Urban	Rural	Eastern	Central	Western	
NTDs	1.6(1.5–1.7)	4.0(3.9–4.2)	1.5(1.4–1.6)	2.8(2.6–2.9)	3.9(3.8–4.1)	2.7(2.7–2.8)
Anencephalus	0.4(0.3–0.4)	1.0(1.0–1.1)	0.4(0.3–0.4)	0.5(0.5–0.6)	1.1(1.1–1.2)	0.7(0.6–0.7)
Spina Bifida	1.0(1.0–1.1)	2.4(2.3–2.5)	1.0(0.9–1.1)	1.9(1.8–2.0)	2.1(2.0–2.3)	1.7(1.6–1.7)
Encephalocele	0.2(0.2–0.3)	0.6(0.5–0.6)	0.2(0.2–0.2)	0.4(0.3–0.4)	0.6(0.6–0.7)	0.4(0.4–0.4)
Hydrocephalus	4.6(4.5–4.8)	5.7(5.5–5.9)	4.4(4.2–4.6)	5.4(5.3–5.6)	5.5(5.3–5.7)	5.1(5.0–5.2)
Anotia/microtia	3.2(3.1–3.3)	2.5(2.4–2.6)	2.9(2.8–3.0)	2.3(2.2–2.4)	3.5(3.3–3.7)	2.9(2.8–2.9)
CHDs	71.8(71.3–72.4)	44.2(43.8–44.7)	74.3(73.6–75.0)	50.2(49.7–50.8)	54.3(53.7–54.9)	59.2(58.9–59.6)
TGA	0.9(0.9–1.0)	0.6(0.6–0.7)	1.3(1.2–1.4)	0.7(0.6–0.8)	0.4(0.4–0.5)	0.8(0.8–0.9)
VSD	14.2(14.0–14.4)	8.6(8.4–8.8)	17.2(16.8–17.5)	9.3(9.1–9.5)	8.8(8.5–9.0)	11.7(11.5–11.8)
ASD	45.5(45.1–46.0)	27.3(27.0–27.7)	44.7(44.2–45.3)	32.6(32.1–33.0)	35.0(34.5–35.5)	37.2(36.9–37.5)
AVSD	2.0(1.9–2.1)	1.3(1.3–1.4)	1.4(1.3–1.5)	1.4(1.3–1.5)	2.4(2.2–2.5)	1.7(1.6–1.8)
TOF	1.5(1.4–1.6)	1.1(1.1–1.2)	1.7(1.6–1.9)	1.4(1.3–1.5)	0.9(0.8–0.9)	1.3(1.3–1.4)
PDA	25.3(25.0–25.6)	14.6(14.4–14.9)	28.1(27.7–28.5)	15.8(15.5–16.1)	18.1(17.7–18.4)	20.4(20.2–20.6)
CP	3.0(2.9–3.1)	2.2(2.1–2.3)	3.4(3.3–3.6)	2.2(2.1–2.3)	2.3(2.2–2.4)	2.6(2.6–2.7)
CL/P	6.3(6.1–6.5)	10.5(10.3–10.7)	6.4(6.2–6.6)	8.6(8.4–8.8)	9.7(9.4–9.9)	8.2(8.1–8.4)
CL	2.8(2.7–2.9)	4.0(3.8–4.1)	2.8(2.7–3.0)	3.4(3.2–3.5)	3.9(3.7–4.1)	3.3(3.3–3.4)
CLP	3.5(3.4–3.6)	6.5(6.4–6.7)	3.5(3.4–3.7)	5.2(5.1–5.4)	5.8(5.6–6.0)	4.9(4.8–5.0)
Anorectal atresia/stenosis	2.9(2.8–3.1)	2.7(2.6–2.8)	3.0(2.8–3.1)	2.8(2.7–3.0)	2.7(2.5–2.8)	2.8(2.8–2.9)
Hypospadias§	6.3(6.1–6.4)	3.6(3.5–3.8)	6.7(6.5–6.9)	4.3(4.1–4.4)	4.4(4.2–4.6)	5.1(5.0–5.2)
Club foot	5.7(5.6–5.9)	6.2(6.1–6.4)	6.2(6.0–6.4)	5.1(4.9–5.3)	6.7(6.5–6.9)	5.9(5.8–6.1)
Polydactyly	18.9(18.6–19.2)	15.8(15.6–16.1)	18.0(17.6–18.3)	16.4(16.1–16.7)	18.3(18.0–18.7)	17.5(17.3–17.7)
Syndactyly	6.7(6.5–6.8)	4.4(4.3–4.6)	6.8(6.6–7.0)	5.1(5.0–5.3)	5.1(4.9–5.3)	5.7(5.5–5.8)
LRD	3.1(3.0–3.2)	3.6(3.5–3.7)	2.9(2.8–3.1)	3.4(3.2–3.5)	3.6(3.5–3.8)	3.3(3.2–3.4)
Omphalocele†	1.0(1.0–1.1)	1.0(0.9–1.0)	0.9(0.9–1.0)	1.0(0.9–1.0)	1.1(1.0–1.2)	1.0(0.9–1.0)
Gastroschisis	0.5(0.5–0.5)	1.2(1.1–1.2)	0.5(0.5–0.6)	0.9(0.8–0.9)	1.0(0.9–1.1)	0.8(0.8–0.8)
Down syndrome	1.7(1.7–1.8)	1.1(1.1–1.2)	1.7(1.6–1.8)	1.2(1.1–1.3)	1.6(1.5–1.7)	1.5(1.4–1.5)

Abbreviations: CI=confidence interval; NTDs=neural tube defects; CHDs=congenital heart diseases; TGA=transposition of great arteries; VSD=ventricular septal defect; ASD=atrial septal defect; AVSD=atrioventricular septal defect; TOF=tetralogy of Fallot; PDA=patent ductus arteriosus; CP=cleft palate; CL/P=cleft lip with or without palate; CL=cleft lip without palate; CLP=cleft lip with palate; LRD=limb reduction defects.

* Per 10,000 perinatal births.

† Except omphalocele, all selected major birth defects varied significantly by urban-rural areas ($p<0.01$); All selected major birth defects varied significantly by geographic location ($p<0.05$ for anorectal atresia/stenosis and omphalocele; $p<0.01$ for the rest).

§ The prevalence rate of hypospadias was defined as the number of case per 10,000 male perinatal births during the specified period.

the prevalence by maternal age groups, a gradual U-shape was identified for NTDs, hydrocephalus, anotia/microtia, CL/P, club foot, polydactyly, syndactyly, LRD, anorectal atresia/stenosis and omphalocele. A higher prevalence in males was found for hydrocephalus, anotia/microtia, CHDs, CL/P, anorectal atresia/stenosis, club foot, polydactyly, syndactyly, LRD, and Down syndrome, while a higher prevalence for females was observed for NTDs and CP.

Significant urban/rural differences in prevalence were identified for several defects. The prevalence of anotia/microtia, CHDs, CP, anorectal atresia/stenosis, hypospadias, polydactyly, syndactyly, and Down syndrome appeared higher in urban areas than in rural areas, while the prevalence of NTDs, hydrocephalus, CL/P, club foot, LRD, and gastroschisis were higher in rural areas. With respect to a geographic disparity in prevalence, the highest prevalence of CHDs, CP, anorectal atresia/stenosis, hypospadias, and syndactyly, and Down syndrome were found in the eastern region,

whereas the highest rates of NTDs, hydrocephalus, CL/P, anotia/microtia, club foot, polydactyly, LRD, omphalocele, and gastroschisis were in the western region.

Figure 1 illustrated the time trends in the prevalence of selected BDs. The prevalence of NTDs, hydrocephalus, CL/P, LRD, omphalocele and gastroschisis decreased during 2010–2018. The defect with the largest decline in prevalence was gastroschisis, followed by NTDs and CL/P, with a decline of 78.8%, 77.3%, and 56.4%, respectively. The prevalence of CHDs, CP, polydactyly, syndactyly, hypospadias, and club foot increased. Compared with rates in 2010, the prevalence of CHDs, syndactyly, and polydactyly in 2018 increased by 171.4%, 60.7%, and 41.2%, respectively. Notably, the prevalence of anotia/microtia, anorectal atresia/stenosis, and Down syndrome remained stable over time.

DISCUSSION

Using 2010–2018 CBDMN data of over 18 million births, we described the epidemiological pattern of 15 selected major BDs at the national level, with special interests in trends over time and perinatal prevalence by maternal age, maternal residence, infant sex, and geographic region. These data will help to clarify the current epidemiological distributions of the top ten most frequent congenital anomalies in the Chinese population and will help determine the disorders and populations that should be prioritized for prevention.

Compared with previous CBDMN data, the overall

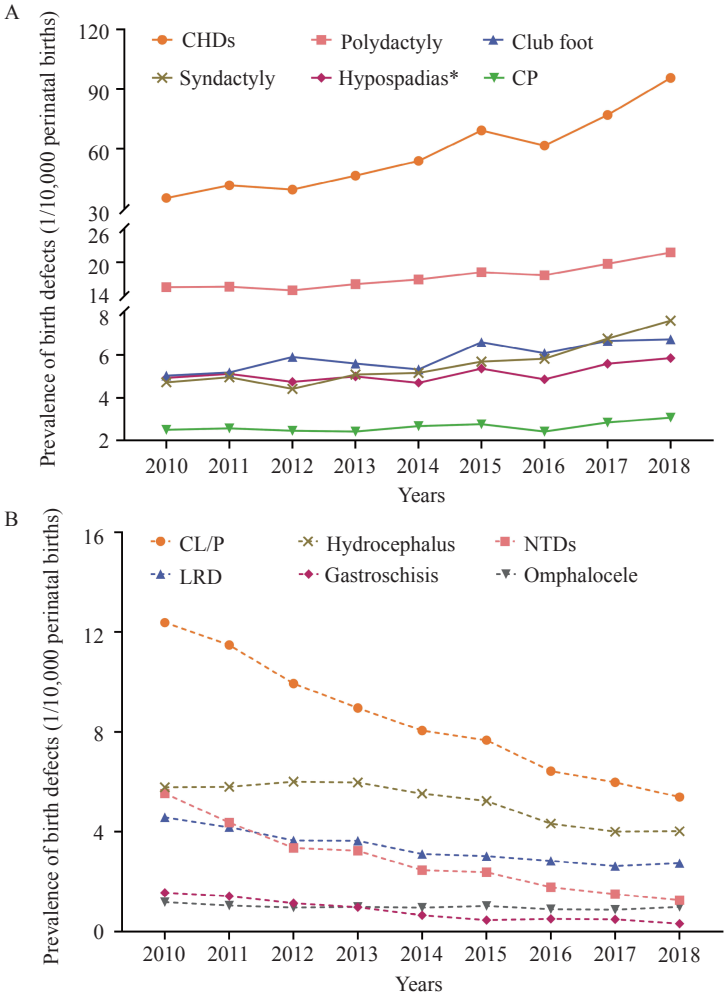

FIGURE 1. Prevalence (1/10,000 perinatal births) of selected major birth defects in China, 2010–2018. (A) Significant increasing trends in the prevalence of six types of birth defects; (B) Significant decreasing trends in the prevalence of six groups of birth defects. * The prevalence of hypospadias was defined as the number of case per 10,000 male perinatal births during the specified period. Abbreviations: CHDs=congenital heart diseases; CP=cleft palate; CL/P=cleft lip with or without palate; NTDs=neural tube defects; LRD=limb reduction defects.

prevalence of CHDs, polydactyly, syndactyly, CP and hypospadias were higher, while the rates of NTDs, hydrocephalus, LRDs, gastroschisis and Down syndrome were lower (*2*,*4*,*6*–*8*). Similar results have been noted in several recent studies in China based on provincial or local hospital-based surveillance data (*9*–*10*). Our prevalence data on CHDs, CL/P, hydrocephalus, hypospadias, anorectal atresia/stenosis and CP, were also comparable to those from hospital-based monitoring systems in Israel, Japan, and Spain (*11*). Notably, the prevalence of CHDs, CL/P, and anotia/microtia in China were higher than those in the United States and some European countries, but Down syndrome prevalence was significantly lower (*11*–*13*). The considerable variations among studies can be explained by ethnicity, socioeconomic factors, environmental exposures, lifestyle risk factors and heterogeneities of surveillance systems or study designs such as differences in inclusion and exclusion criteria, diagnostic capability, and follow-up time.

A growing number of epidemiological studies reveal that greater risks or higher prevalence of certain BDs happen in younger or older women. In the current analysis, we identified an U-shaped pattern for 10 types of anomalies (NTDs, CL/P, etc., Table 1), and increased prevalence of 4 defects with maternal age (CHDs, Down syndrome, CP, and hypospadias). Maternal-age-specific prevalence patterns varied by the types of BDs, which might be partially due to the changing maternal age distribution following by the implementation of the two-child policy (*9*). In our study, significant urban-rural and geographic disparities were found for nearly all the selected BDs, which could be explained by differences in occupational exposures, socioeconomic levels, and healthcare among people living in different area types and regions. Similar findings have been noted in other reports, indicating considerable health inequalities in China (*4*,*6*–*8*,*10*). Therefore, BDs prevention and healthcare services in rural and western regions need to be further improved (*14*).

Consistent with previous studies in China, an increasing prevalence for CHDs, CP, polydactyly, syndactyly, and hypospadias during 2010–2018 was found (*2*,*9*–*10*). Indeed, the increased overall CHD prevalence can be largely attributed to substantial increases in several mild lesions (i.e. small ventricular septal defect, atrial septal defect, and patent ductus arteriosus, etc.). Improvements in diagnostic capabilities, disease screening, and the widespread use of echocardiography can lead to the earlier identification of mild lesions or asymptomatic CHD subtypes and lead to a higher detection rate of CHDs in the perinatal period (*9*–*10*,*12*–*13*). Changes in environmental exposures like exogenous estrogenic endocrine disruptors have been reported to be associated with increased hypospadias prevalence (*10*,*13*), but the exact reasons for increased prevalence of CP, polydactyly, and syndactyly were unclear. The decreasing prevalence of NTDs, hydrocephalus, LRD, and gastroschisis might reflect the combined effect of strengthened primary and secondary prevention (i.e. the National Folic Acid Supplementation Program and prenatal screening and diagnosis for structural malformations and Down syndrome) in China (*2*,*9*–*10*). Given the high prevalence of these defects, we believe further etiological studies are needed, and postnatal care, surgical correction, rehabilitation, and social support should be strengthened.

This study was subject to some limitations. The calculation of perinatal prevalence rate in the current analysis excluded cases <28 weeks of gestation. The rates mainly reflected the effect of combinations of risk factors and primary and secondary preventions and could not be simply explained as an indicator of disease risk. Hospital-based CBDMN data may introduce referral bias, but the impact could be minimal because of the high hospital delivery rate in China (≥99.9%) (*15*). Since the follow-up time period was relatively short (28 weeks of gestation to Day 7 after birth), CBDMN had limited ability to obtain reliable data on congenital metabolic diseases, functional abnormalities, and outcomes in the infancy period. However, considering the large sample size and wide geographic coverage, this data can well represent the epidemiological characteristics of most structural malformations in China.

In summary, we presented the prevalence patterns of 15 major BDs during 2010–2018, mainly focusing on the time trends and the prevalence of the top ten most frequently occurring structural malformations by maternal and infant characteristics. These findings will contribute to health policy making and future BDs prevention by providing important baseline references.

Acknowledgements: We thank all the colleagues from local institutions for participating in the data collection.

Conflicts of interest: The authors declare no competing interests.

doi: 10.46234/ccdcw2020.195

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Submitted: August 25, 2020; Accepted: September 09, 2020

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