Rachel's Environment & Health News
#410 - Birth Defects -- Part 1
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The Birth Defects Monitoring Program (BDMP) is a U.S. government effort to monitor birth defects (congenital malformations) using data collected when newborn infants are discharged from the hospital.[1] The BDMP was initiated by the federal Centers for Disease Control (CDC) in 1974. The current BDMP database includes information on roughly 15 million births that have occurred at 1200 predominantly mid-sized community hospitals in the U.S. during the past 20 years.

The BDMP database is not comprehensive (it does not include information on every birth that occurs in the U.S.). Neither does it represent a randomly-selected sample of all U.S. births; therefore data from the BDMP cannot be considered representative of the entire "universe" of all U.S. newborns. In 1987 the BDMP received information on 15% of all U.S. births, which gives an idea of how comprehensive the coverage is. Because the data are mostly from mid-sized hospitals, we might expect that some of the largest hospitals in the largest cities are under- represented. Nevertheless, as the CDC says, the BDMP "represents the largest single set of uniformly collected and coded discharge data on congenital malformations in the United States." It is simply the best information available on birth defects in the U.S.

CDC says that the BDMP "functions primarily as an early warning system; however it can be useful also for correlating incidence [occurrence] patterns with such trends as the temporal [time-related] and geographic distribution of drugs, chemicals, and other possible human teratogens." A teratogen (from the Greek words meaning "monster producing") is anything that causes birth defects. Examples of teratogens are diseases such as German measles; infections; inherited genetic defects; radiation; and certain chemicals.

In 1990, researchers looked for trends in the BDMP database, examining records for 38 types of birth defects from 1979-80 through 1986-87. During this 7-year period, of the 38 types of birth defects, 29 increased; 2 decreased; and 7 remained stable (meaning they changed less than 2% per year during the 7-year period.)

Table 1 shows the annual percent change for 30 types of birth defects. All of them increased during the 7-year period (though some increased at a rate less than 2% per year, and are thus classified as "stable" by the CDC).

Table 1 contains 3 columns of numbers. The first two columns show the actual number of birth defects per 10,000 births; the first column shows data for the earlier period, 1979-80; the second column shows the later period, 1986-87. The third column shows the yearly percentage increase during the 7-year period.

Some of these increases are explained by better health care and better diagnosis. For example, some of the heart defects listed in Table 1 are so serious that an infant might not have survived such a defect 10 years ago but might survive it today. Likewise, some of the heart defects might be revealed by high-tech medical diagnostic machines today, whereas they might have gone unnoticed 10 years ago.

However, many of the increases in birth defects in Table 1 cannot be explained by better health care or better diagnosis. If a child were born 10 years ago with the iris missing from one or both of its eyes, chances are good that the mother or her doctor or a nurse would see it. (The iris is the part of the eye that makes blue eyes blue and brown eyes brown.) So the 5.2% ANNUAL INCREASE in "aniridia" (absence of an iris) is very likely a real increase.

The same can be said for birth defects of the central nervous system, facial clefts, musculoskeletal defects and some of the gastrointestinal and genitourinary defects. Most of these defects are so obvious that they would have been noticed as easily 10 years ago as today. Therefore, increases in these defects are very likely real increases.

Some of the increases shown in Table 1 are surprisingly large. For example, coloboma of the eye increased 9.6% each year during the 7-year period; this means the occurrence of this defect doubled during the study period. (Coloboma of the eye means a wedge-shaped piece is missing from the iris, or some other part of the eye is missing.) Other eye disorders (congenital cataract, for example) are increasing about 5% each year, thus doubling every 14 years. (The relation-ship of annually-increasing quantities to the doubling time was detailed in REHW #197 and #199.)

Are most birth defects caused by the parents' genetic characteristics, or by something in the environment?

In July of this year an important study of birth defects in Norway appeared in the NEW ENGLAND JOURNAL OF MEDICINE.[2] It indicated that environmental factors may be more important than previously thought.

Norway has maintained a Medical Birth Registry since 1967; the registry now contains data on 1.5 million births. Norwegian and American researchers examined records of 371,933 women who had given birth to first and second children in Norway between 1967 and 1989. For the 9192 women whose first infant had a birth defect, they examined the risk of similar or dissimilar effects in the second infant. And they examined the risk of a birth defect in the second child among mothers who lived in the same municipality during both pregnancies vs. mothers who moved to a new municipality before the second child was born. (The control group was the 362,741 women whose first infant did not have a birth defect.)

The researchers found that 2.5% of all infants born in Norway have a birth defect. Examining 23 different kinds of birth defects, they found that in every category, mothers whose first infant had a defect were more likely to have a second infant with a defect, as would be expected if birth defects are genetic in origin. What was "surprising" to the researchers was that women who moved to a new city between pregnancies were only half as likely to have a second child with a birth defect. Mothers whose first child had a defect were 11.6 times as likely to have a second child with a defect (compared to mothers whose first child did not have a defect), but if a mother moved to a new municipality between pregnancies she was only 5.1 times as likely to have a second child with a defect. The researchers concluded, "...[W]e find strong, if indirect, evidence... suggesting that important environmental teratogens have yet to be discovered."

--Peter Montague

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[1] Larry D. Edmonds, "Temporal Trends in the Prevalence of Congenital Malformations at Birth Based on the Birth Defects Monitoring Program, United States, 1979-1987," MORBIDITY AND MORTALITY WEEKLY REPORT, CDC SURVEILLANCE SUMMARIES Vol. 39, No. SS-4 (Dec., 1990), pgs. 19-23.

[2] Rolv Terje Lie and others, "A Population-Based Study of the Risk of Recurrence of Birth Defects," NEW ENGLAND JOURNAL OF MEDICINE Vol. 331, No. 1 (July 7, 1994), pgs. 1-4.

===== . TABLE 1 Birth Defects: Annual Percent Change in Occurrence During 7-year Period, 1979-80 to 1986-87, United States ===== . Annual . Number of Percent . defects per change in DEFECT TYPE 10,000 births occurrence, . ---------- 1979-80 to . 1979-80 1986-87 1986-87 CENTRAL NERVOUS SYSTEM Hydrocephalus without spina bifida . (fluid in the skull) 4.34 5.84 4.3% Encephalocele (gap in the skull) 1.10 1.16 0.8% Microcephalus (small head) 2.12 2.61 3.0% . EYES Anophthalmos (absence of eyes) 0.57 0.68 2.6% Congenital cataract . (eye cataracts at birth) 0.71 1.02 5.3% Coloboma of eye

(eye parts missing) 0.21 0.40 9.6% Aniridia (absence of the iris) 0.07 0.10 5.2% . HEART Common truncus . (undeveloped main arteries)9 0.19 0.40 11.2% Transposition of great arteries . (reversal of main arteries) 0.87 1.45 7.6% Tetralogy of Fallot . (4 common defects simultaneously) 0.73 1.82 13.9% Ventricular septal defect . (opening between lower chambers) 11.34 20.49 8.8% Atrial septal defect . (opening between upper chambers) 1.16 3.69 18.0% Endocardial cushion defect 0.34 0.95 15.8% Pulmonary valve atresia and stenosis . (obstructed blood flow) 0.58 3.44 29.0% Tricuspid valve atresia and stenosis . (obstructed blood flow) 0.16 0.36 12.3% Aortic valve stenosis and atresia . (obstructed blood flow) 0.22 0.79 20.0% Hypoplastic left heart syndrome . (undeveloped left side) 0.56 1.25 12.2% Patent ductus arteriosus . (pulmonary artery open to aorta) 17.87 35.43 10.3% Coarctation of aorta . (constriction of the aorta) 0.74 1.15 6.5% Pulmonary artery anomaly 1.12 2.66 13.2% Lung agenesis and hypoplasia . (undeveloped lungs) 1.66 3.84 12.7% . FACIAL CLEFTS Cleft palate without cleft lip 5.05 5.33 0.8% Cleft lip 7.76 9.35 2.7% . GASTROINTESTINAL Tracheoesophageal anomalies . (upper airway problems) 1.86 2.49 4.3% Rectal and intestinal atresia . (blockage) 3.23 3.80 2.3% . GENITOURINARY Renal agenesis and hypoplasia . (one kidney or small kidneys) 1.23 2.34 9.6% Bladder exstrophy . (gap in abdomen, . revealing bladder) 0.29 0.33 1.9% . MUSCULOSKELETAL Reduction deformity, upper limbs . (arms deformed or missing) 1.53 1.58 0.5% Reduction deformity, lower limbs . (legs deformed or missing) 0.78 0.83 0.9% Congenital arthrogryposis . (contracted or bent limbs) 1.33 1.93 5.5% . ========== Source: Larry D. Edmonds and others, "Temporal Trends in the Prevalence of Congenital Malformations at Birth Based on the Birth Defects Monitoring Program, United States, 1979-1987," MORBIDITY AND MORTALITY WEEKLY REPORT, CDC SURVEILLANCE SUMMARIES Vol. 39, No. SS-4 (December, 1990), pg. 22.

Descriptor terms: birth defects monitoring program; cdc; new england journal of medicine; norway; hydrocephalus; encephalocele; microcephalus; anophthalmos; congenital cataract; coloboma of the eye; aniridia; common truncus; heart; head; brain; tetralogy of fallot; ventricular septal defect; atrial septal defect; endocardial cushion defect; pulmonary valve atresia and stenosis; tricuspid valve atresia and stenosis; aortic valve stenosis and atresia; hypoplastic left heart syndrome; patent ductus arteriosus; coarctation of aorta; pulmonary artery anomaly; lung agenesis and hypoplasia; cleft palate; tracheoesophageal anomalies; rectal and intestinal atresia; renal agenesis and hypoplasia; reduction deformity; congenital arthrogryposis;