

Birth Defects

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Criteria

- **Substantial public health burden**
- **Well-defined case definition**
- **Awareness of disease among relatives**
- **Accurately reported by family members**
- **Family history is established risk factor**
- **Effective interventions for primary and secondary prevention**

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Case Definition

- Birth Defect?
- *A birth defect is an abnormality of **structure** or **function** present at birth that results in physical disability, mental disability or death*

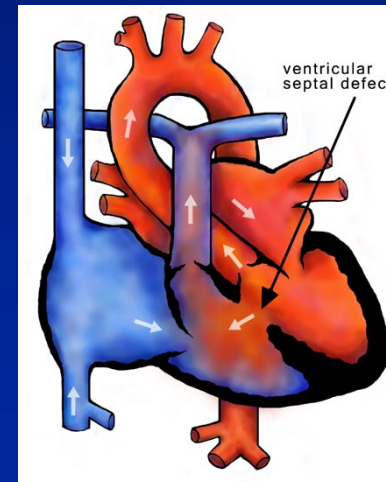
Case Definition

- **Structural Defects**

- Brain/Spinal Cord
- Ear
- Eye
- Facial/Oral
- Gastrointestinal
- Genital/Urinary
- Heart
- Muscle/Skeletal



Cleft lip and palate



Ventricular septal defect

Child photo courtesy of NFFR

Cleft Lip +/- Palate

- **DEFINITION -- incomplete closure of the lip; often accompanied by a maxillary alveolar (gum) defect and/or cleft palate ; maxillary alveolar defect may be a complete cleft that is continuous with the cleft palate, or it may be limited to a notch on the gum; cleft lip may be unilateral, bilateral, or median (distinguished from bilateral cleft lip by agenesis of premaxilla)**

Cleft Lip +/- Palate

- **COMPLETE CLEFT LIP**--defect extends through the entirety of the lip and the nasal floor; may be unilateral or bilateral; usually associated with a more severe nasal deformation
- **INCOMPLETE CLEFT LIP**--defect of lip that does not extend into the nasal floor; may be unilateral or bilateral; there may be an incomplete cleft lip on one side and a complete cleft lip on the other side
- **PSEUDOCLEFT LIP**--abnormal linear thickening or depressed groove of skin, or subtle scar-like pigmentary difference paralleling the philtral ridge on the affected side; may be associated with slight notch of the vermillion or a mild slouching of the alar cartilage

Source: CDC

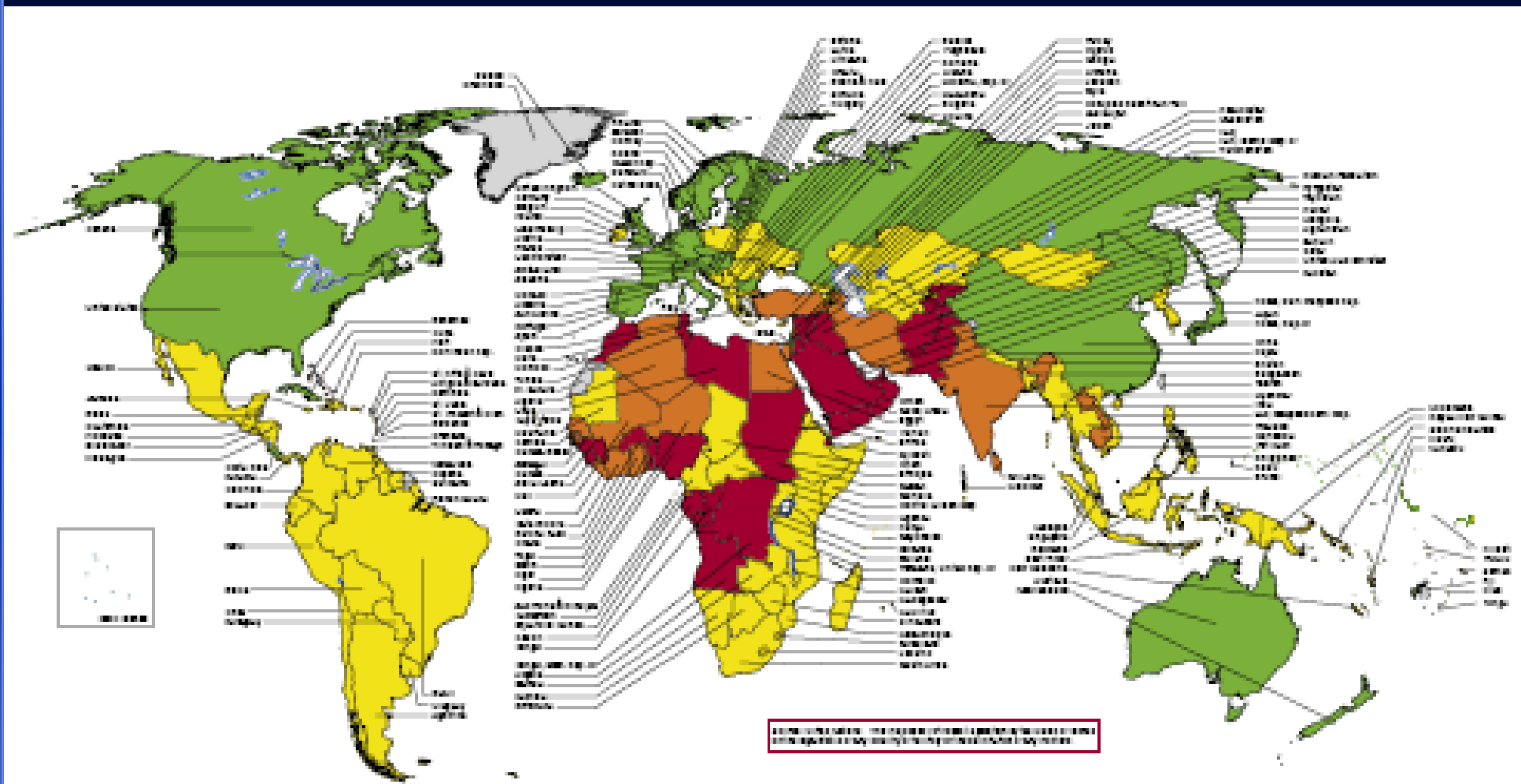
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Public Health Burden – World

- Overall estimated 6% of births have defect of genetic or partially genetic origin
- Heart defects, neural tube defects and Down syndrome most common defects
- Estimated 3.3 million children under 5 years of age die annually from serious birth defects
- Highest totals of occurrence (94%) and deaths (95%) found in middle- and low-income countries

Source: March of Dimes 2006



Source: March of Dimes 2006

Public Health Burden – US

- Overall estimated birth prevalence of 3-5%
- National estimates of 18 selected defects found highest prevalence for orofacial clefts and Down syndrome*
- Leading cause of infant mortality (1 in 5 deaths)
- On average, 18 babies die per day as result of birth defect
- Costs for care and treatment annually totals millions of dollars

*Source: MMWR 2006

Public Health Burden

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Established Risk Factor

- **20-30% due to known genetic factors**
- **10% due to known environmental factors**
- **60-70% due to unknown genetic and/or environmental factors**

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Awareness of Birth Defects

**Facial
Cleft**



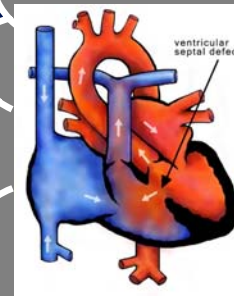
Child photo courtesy of NFFR

Awareness of Birth Defects

- Facial cleft?
 - Cleft lip
 - Cleft palate
 - Harelip
 - Bifid uvula
 - Hole in palate

Awareness of Birth Defects

Heart
Defect



Awareness of Birth Defects

- Heart defect?
 - Ventricular septal defect
 - Heart murmur
 - Heart on wrong side
 - Hole in the heart
 - Myxoma

Awareness of Birth Defects

Birth
Defect or
Condition

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Awareness of Birth Defects

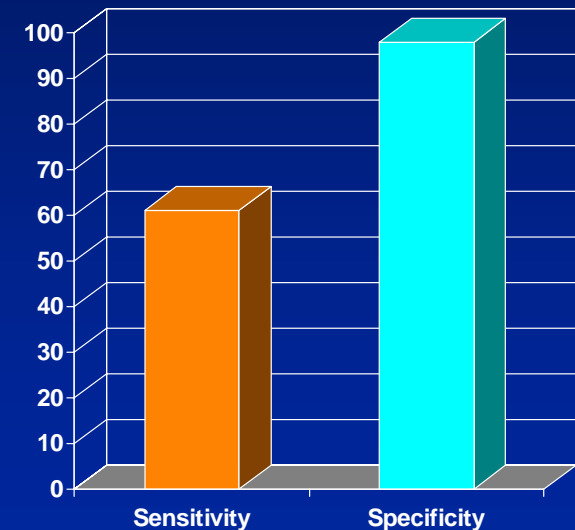
- Birth defect or condition?
- Achondroplasia
- Arthritis
- Crossed eyes
- Cerebral palsy
- Left handed

Accurately Reported

Goal: Evaluate the quality of case and control mother interview reports of birth defects among offspring



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Compared reports with data from Metropolitan Atlanta Congenital Defects Program

Source: Rasmussen et al., 1989

Accurately Reported

Goal: Evaluate quality of case and control mother interview reports of birth defects (and cancer) among offspring and first-, second- and third-degree relatives



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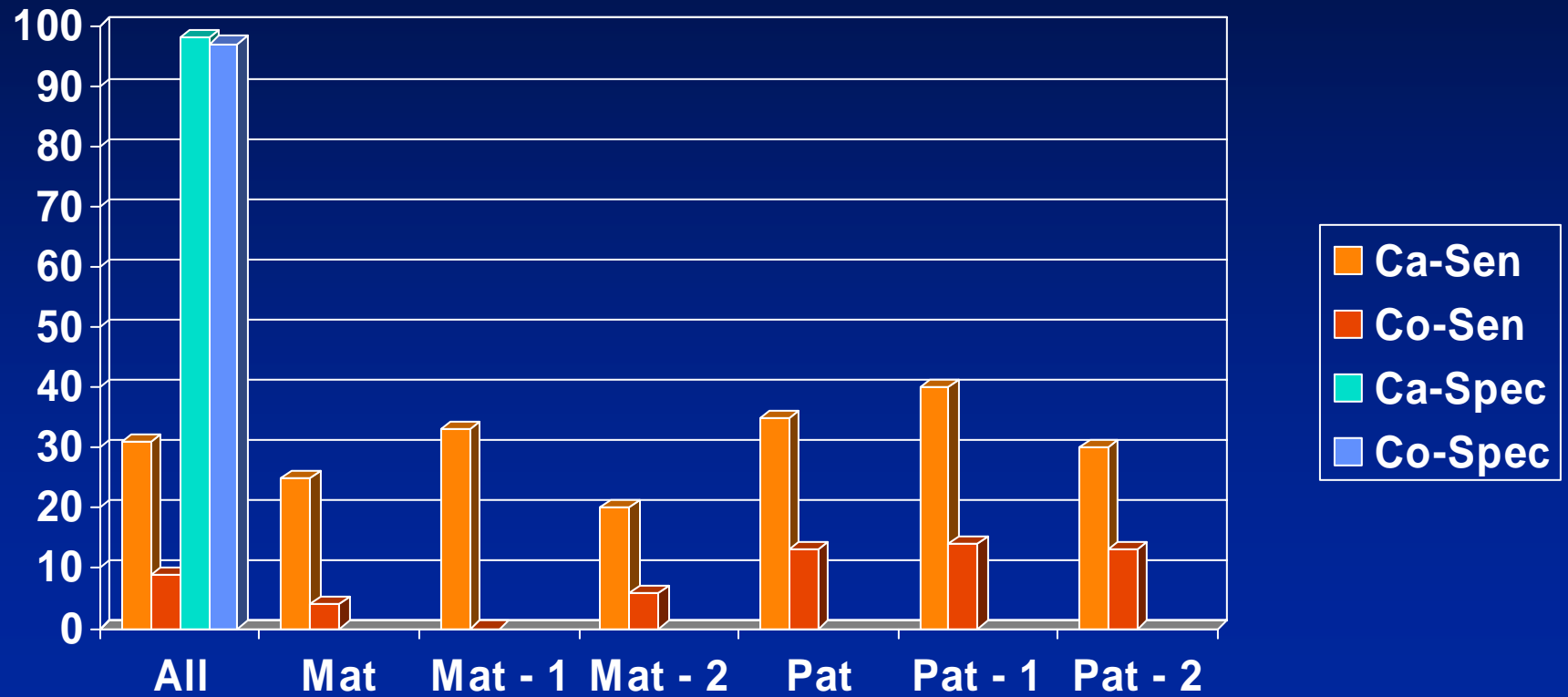


**Compared maternal reports
with relative self-reports**

**Compared reports with
Registry data**

Source: Romitti et al., 1997

Accurately Reported



Source: Romitti et al., 1997

Accurately Reported

Family

- Number of relatives
- Gender of relatives
- Index child birth order

Maternal

- Age
- Education
- Family genealogy
- Social contact



Investigated effects of selected family and maternal characteristics on quality of reports

Accurately Reported

- Sensitivity of maternal reports differed by study group (ca>co) and type of relative (pat>mat)
- Specificity high for both study groups and each type of relative
- Mothers (ca and co) tended to over-report ear, face and neck defects and eye (ca) and genitourinary defects (co)
- Case status and participation in family genealogy were strongest predictors of concordance

Preliminary Recommendations

- **Self-administered questionnaire**
- **Modular design**
- **Specific, closed-ended items**
- **Systematic inquiries about family members**

Orofacial Clefts

- **Applied recommendations to two case-control studies**
- **Ongoing Iowa-based study**
- **Three-center pilot study (IA, AR, NY) in National Birth Defects Prevention Study**

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Primary Prevention

- **Nutrition**

- Daily intake of multivitamin with folic acid
- Healthy, balanced diet

- **Behaviors**

- Avoid alcohol and tobacco
- Avoid illicit drugs

- **Medical/prenatal care**

- Pre-pregnancy planning including regular medical check-ups
- Medication use (over-the-counter and prescription)

Primary Prevention

- **Family history data collection**
 - Tool for pediatric care
 - Tool for reproductive counseling
- **Recurrence risks***
 - Five- to seven-fold risk for same defect in second child
 - Smaller but elevated risks for different defect in second child
 - More common recurring defects included orofacial clefts and central nervous system, limb and genitourinary defects

*Sources: Lie et al., 1994; Basso et al., 1999

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