

Report
National Birth Defect Registry
April 2009

The National Birth Defect Registry was started in 1990 to collect information on all kinds of birth defects, both structural and functional, as well as information on the preconceptional and prenatal health, genetic and exposure histories of both parents. The project was designed and evaluated by an advisory board of seven prominent scientists working in fields ranging from obstetrics and genetics to epidemiology and the effects of toxic exposures on reproduction. The original questionnaire for the project was a 16-page booklet that parents filled out and mailed to Birth Defect Research for Children. Data from the questionnaire were then manually entered into an interrelational data base. Approximately 4,000 case reports were collected in this version of the registry.

In April of 2004, BDRC converted the registry to an on-line project so that parents could fill out the questionnaire at any convenient time of the day or night. The on-line project also allowed for the addition of new sections to the registry questionnaire without the cost of reprinting thousands of questionnaire booklets and mailing them to parents. The quality of data collected is more accurate as well, since the parent enters information directly into the on-line questionnaire and then BDRC staff imports the questionnaire into the main registry taking the completed questionnaire off the internet.

The registry is designed to identify patterns of similar birth defects that may have similar conditions or exposures in common in the pregnancy or pre-conceptual history. This slide presentation made to the National Institute of Environmental Health Sciences explains the operation of the registry and some of the birth defect issues we have worked on using registry data. <http://www.birthdefects.org/NIEHS/3510.html>

Since April of 2004, parents have completed 1853 questionnaires with case reports for 2030 children (more than one child in a family can be reported on a questionnaire) in the on-line National Birth Defect Registry. We have reports for 1095 males, 926 females and 9 cases where the sex was not entered. This is a first report of these data.

Distribution of Disabilities With a Minimum of 2 Case Reports

| CountOfBirth_Defect_ID | Birth_Defect_Name |
|------------------------|-------------------------|
| 490 | Motor Skills Delay |
| 443 | Speech Delay |
| 386 | Reflux |
| 338 | Allergies |
| 338 | Speech |
| 264 | Frequent Ear Infections |
| 254 | Asthma |

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| 225 | Frequent Upper Respiratory Infections |
| 209 | Eczema |
| 183 | Food Sensitivity |
| 177 | Colic As Baby |
| 175 | Comprehension |
| 173 | Heart Murmur |
| 172 | Malformed Fingers, Hands Or Arms |
| 167 | Spelling/Writing |
| 166 | Jaundice At Birth |
| 164 | Scoliosis |
| 159 | Language Processing |
| 157 | Cleft Palate |
| 156 | Sensory Integration Dysfunction |
| 152 | Math LD |
| 151 | Attention Deficit With Hyperactivity |
| 149 | Anger |
| 148 | Drug Reaction |
| 147 | Mood Swings |
| 143 | Retardation |
| 139 | Anxiety Disorder |
| 138 | Unexplained Fevers |
| 137 | Muscle Pain/Weakness |
| 136 | Hypotonia |
| 134 | Abnormal Teeth |
| 134 | Chronic Stomach Problems |
| 134 | Frequent Headaches |

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| 134 | Seizure Disorder |
| 133 | Sleep Disorder |
| 127 | Joint Pain |
| 125 | Partial Hearing Loss |
| 122 | Distractibility |
| 120 | Depression |
| 120 | Memory Problems |
| 119 | Ventricular Septal Defect |
| 113 | High Arched Palate |
| 112 | Heat/Cold Sensitivity |
| 112 | Malformed Toes, Feet Or Legs |
| 111 | Clubfoot |
| 107 | Gastroschisis |
| 105 | External Ear Or Ears (Absent or Malformed) |
| 105 | Low Set Ears |
| 105 | Strawberry Birth Mark |
| 97 | Cleft Lip |
| 97 | Hydrocephaly |
| 97 | Obsessive/Compulsive |
| 97 | Tracheo-Esophageal Anomaly |
| 96 | Facial Asymmetry |
| 96 | Tethered Spinal Cord |
| 95 | Apnea |
| 95 | Latex |
| 94 | Frequent Urinary Tract Infections |
| 94 | Malformations Of Vertebrae |

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|----|---|
| 92 | Autism |
| 91 | Atrial Septal Defect |
| 90 | Lung (Absent Or Underdeveloped) |
| 90 | Vision Defect |
| 87 | Amblyopia (Lazy Eye) |
| 87 | Auditory Processing Deficit |
| 87 | Hayfever |
| 87 | Microcephaly |
| 85 | Undescended Testicles |
| 84 | Spina Bifida |
| 82 | Astigmatism |
| 81 | Cerebral Palsy |
| 79 | Light Sensitivity |
| 79 | Muscle (Absent Or Underdeveloped) |
| 78 | Imperforate Anus |
| 77 | Arnold Chiari Malformation |
| 77 | Walks On Tiptoes |
| 76 | Rib Abnormality |
| 75 | Attention Deficit Without Hyperactivity |
| 73 | Anemia |
| 73 | Dyslexia/Reading |
| 73 | Frequent Pneumonia |
| 71 | Syndactly (Webbed Fingers Or Toes) |
| 70 | Diaphragmatic Hernia |
| 69 | In Utero Growth Retardation |
| 67 | Strabismus |

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|----|--|
| 65 | Hives |
| 64 | Acne-Like Rash |
| 64 | Chronic Fatigue |
| 64 | Micrognathia (Small Jaw) |
| 61 | Goldenhar (Hemifacial Microsomia) |
| 60 | Internal Ear Or Ears (Absent or Malformed) |
| 59 | Menstrual Problems |
| 58 | Absence Corpus Callosum |
| 58 | Dislocated Hip |
| 58 | Vater (Vacterl) |
| 55 | Ear Tag(S) |
| 55 | Patent Ductus Arteriosis |
| 54 | Neurogenic Bladder |
| 52 | Umbilical Hernia |
| 51 | Deafness |
| 51 | Reduction Deformity Upper Limb |
| 50 | Absent Kidney |
| 49 | Dysmorphic Features |
| 49 | Oppositional Defiant Disorder |
| 48 | Frequent Nose Bleeds |
| 47 | PDD (Pervasive Developmental Disorder) |
| 47 | Thyroid Defect |
| 46 | Visual Memory Deficits |
| 45 | Cafe Au Lait Birth Mark |
| 44 | Small Sex Organs |
| 42 | Apraxia |

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|----|--------------------------------|
| 42 | Bony Defect Of Skull |
| 41 | Chromosome Disorder |
| 41 | Early Puberty |
| 40 | Anencephaly |
| 40 | Constitutional Short Stature |
| 39 | Blind |
| 39 | Hypospadias |
| 38 | Asperger's |
| 37 | Growth Hormone Deficiency |
| 37 | Sensory Neural Hearing Loss |
| 36 | Craniosynostosis |
| 36 | Hypoplasia Left Heart Syndrome |
| 36 | Perseveration |
| 35 | Facial Bone Deformity |
| 35 | Hypertension |
| 35 | Pectus Excavatum |
| 34 | Early Tooth Eruption |
| 34 | Immune Deficiency |
| 34 | Ovarian Cyst |
| 34 | Poland's |
| 33 | Bi-Polar |
| 33 | Hemangioma |
| 33 | Nystagmus |
| 33 | Spina Bifida Occulta |
| 32 | Arthritis |
| 32 | Neurogenic Bowel |

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|----|---------------------------------------|
| 31 | Malformed Ureter |
| 31 | Reduction Deformity Lower Limb |
| 31 | Thrush/Chronic Candida |
| 30 | Hiatal Hernia |
| 29 | Amniotic Band |
| 29 | Clubfoot With Bone Deformity |
| 29 | Inguinal Hernia |
| 29 | Polydactyly (Extra Digits) |
| 29 | Pulmonary Hypertension |
| 28 | Coarctation Of Aorta |
| 28 | Coloboma |
| 28 | Contractures Of Elbows/Knees/Joints |
| 28 | Hair Loss |
| 28 | Pectoralis (Absent Or Underdeveloped) |
| 28 | Single umbilical artery |
| 28 | Tetralogy Of Fallot |
| 27 | Bronchial Pulmonary Dysplasia |
| 27 | Changes In Skin Color |
| 27 | Holoprosencephaly |
| 27 | Hypoactivity |
| 27 | Hypoglycemia |
| 27 | Macrocephaly |
| 27 | Malformed Kidney |
| 26 | Brain Cyst |
| 26 | Delayed Puberty |
| 26 | Transposition Great Arteries |

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|----|------------------------------------|
| 25 | Facial Nerve Paralysis |
| 25 | Pulmonary Valve Stenosis/Atresia |
| 25 | Rectal And/Or Intestinal Atresia |
| 25 | Swollen/Or Bleeding Gums |
| 24 | Aortic Valve Stenosis/Atresia |
| 24 | Psoriasis |
| 24 | Thin Corpus Callosum |
| 23 | Autoimmune Disorder |
| 23 | Chronic Tinnitus (Ringing In Ears) |
| 23 | Encephalocele |
| 22 | Cataract |
| 22 | Cortical Blindness |
| 22 | Diabetes |
| 22 | Pierre Robin |
| 22 | Pyloric Stenosis |
| 21 | Clubbed Hands |
| 20 | Breaks Law/Rules Of Society |
| 20 | Chronic Virus |
| 20 | Infertility |
| 20 | Malformed Sex Organs |
| 20 | Microphthalmia |
| 20 | Pituitary Defect |
| 19 | Intrauterine Stroke |
| 19 | Lipoma |
| 18 | Bicuspid Aortic Valve |
| 18 | Cardiac Arrhythmia |

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| 18 | Exotropia |
| 18 | Manic Depression |
| 18 | Pigeon Chest |
| 17 | Horseshoe Kidney |
| 17 | Mitral Valve Prolapse |
| 17 | Optic Nerve Atrophy |
| 17 | Polycystic Kidney |
| 17 | Port Wine Stain |
| 17 | Ptosis |
| 17 | Skin Fungus |
| 17 | Twisted Lower Limb |
| 16 | Ataxia |
| 16 | Dermoid Cysts |
| 16 | Endometriosis |
| 16 | Mitral Valve Regurgitation |
| 16 | Omphalocele |
| 15 | Clinodactyly |
| 15 | Hypertelorism |
| 15 | Pulmonary Atresia/Hypoplasia |
| 14 | Bladder Exstrophy |
| 14 | Bradycardia |
| 14 | Cystic Hygroma |
| 14 | Diabetes Insipidus |
| 14 | Short Bowel |
| 13 | Failure to Thrive |
| 13 | Glaucoma |

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|----|-------------------------------|
| 13 | Potter's |
| 13 | Syringomyelia |
| 13 | Turner's |
| 12 | Ambiguous Genitalia |
| 12 | Aphasia |
| 12 | Chronic constipation |
| 12 | Dandy Walker Malformation |
| 12 | Double Outlet Right Ventricle |
| 12 | G-Tube dependant |
| 12 | Liver Function Disorder |
| 12 | Sacral Agenesis |
| 12 | Septo-Optic Dysplasia |
| 12 | Trisomy |
| 11 | Caudal Regression Syndrome |
| 11 | Charge |
| 11 | Choanal Atresia |
| 11 | Downs' |
| 11 | Lipomyelomeningocele |
| 10 | Cloacal Exstrophy |
| 10 | ganglion cyst |
| 10 | Hepatomegaly (Enlarged Liver) |
| 10 | Hirschsprung Disease |
| 10 | Hydrocele |
| 10 | IBS |
| 10 | Lissencephaly |
| 10 | Overgrowth Of Lower Limb |

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|----|----------------------------------|
| 10 | Rheumatoid Arthritis |
| 9 | Absence Of Septum Pellucidum |
| 9 | Absent Bladder |
| 9 | Arthrogyposis |
| 9 | Cardiomyopathy |
| 9 | Delayed Myelination |
| 9 | Dextrocardia |
| 9 | Macrostomia |
| 9 | Preauricular Pits |
| 8 | Borderline Personality Disorder |
| 8 | Edward's |
| 8 | Hypertonia |
| 8 | Neurofibromatosis |
| 8 | Overgrowth (Giant) |
| 8 | Pulmonary Artery Anomaly |
| 7 | Anophthalmia (Missing Eye) |
| 7 | Cardiomegaly |
| 7 | Ehlers Danlos |
| 7 | hydronephrosis |
| 7 | Hypoplastic Right Heart Syndrome |
| 7 | Lymphangioma |
| 7 | Mitral Valve Stenosis |
| 7 | Periventricular Leukomalacia |
| 7 | Tricuspid Valve Stenosis/Atresia |
| 7 | Vascular Ring |
| 6 | Anorexia Or Bulimia |

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| 6 | Bicornuate Uterus |
| 6 | Cerebellar Hypoplasia |
| 6 | Crohn's Disease |
| 6 | Ebstein's Anomaly |
| 6 | Epispadias |
| 6 | Osteogenesis Imperfecta |
| 6 | Overgrowth Of Upper Limb |
| 6 | Twisted Upper Limb |
| 5 | Adams oliver syndrome |
| 5 | Biliary Atresia |
| 5 | developmental delay |
| 5 | Digeorge |
| 5 | Endocardial Cushion Defect |
| 5 | Hemihypertrophy |
| 5 | Incontinentia Pigmenti |
| 5 | Peripheral Pulmonary Stenosis |
| 5 | Schizencephaly |
| 5 | Splenomegaly (Enlarged Spleen) |
| 5 | Static Encephalopathy |
| 4 | Absent Gall Bladder |
| 4 | Achondroplastic Dwarfism |
| 4 | Aplasia Cutis Congenita |
| 4 | Congenital Cystic Adenomatoid Malformation |
| 4 | Cornelia De Lange |
| 4 | fibular hemimelia |
| 4 | Hydranencephaly |

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| 4 | Leukodystrophy |
| 4 | Macroglossia |
| 4 | Malrotation |
| 4 | Muscular Dystrophy |
| 4 | Red Dye #40 |
| 4 | sacral dimple |
| 4 | Spondylolisthesis |
| 4 | Teratomas |
| 4 | Williams |
| 4 | Wolff-Parkinson White |
| 3 | Albinism |
| 3 | Astrocytoma |
| 3 | cleft mitral valve |
| 3 | CMV Virus |
| 3 | Duane Refraction |
| 3 | Ectodermal Dysplasia |
| 3 | Hernia |
| 3 | kidney reflux |
| 3 | Klippel-Fiel |
| 3 | Lennox-Gastaut |
| 3 | Leukemia |
| 3 | Noonan's |
| 3 | Primary Immune Defect |
| 3 | PTSD |
| 3 | Schizophrenia |
| 3 | Spinal Muscular Atrophy |

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| | 3 | Stickler's |
| | 3 | torticollis |
| | 3 | Truncus Arteriosus |
| | 2 | Absent Spleen |
| | 2 | Acrania |
| | 2 | Adhesives |
| | 2 | Aicardi |
| | 2 | anal stricture |
| | 2 | Auditory Neuropathy |
| | 2 | big forehead |
| | 2 | Bilateral Periventricular Nodular Heterotopia |
| | 2 | Binder Syndrome |
| | 2 | Bladder Spasms |
| | 2 | Cavernous Hemangioma |
| | 2 | Celiac Disease |
| | 2 | cerebellar agenesis |
| | 2 | Chiari malformation |
| | 2 | Chondromalacia Patellae |
| | 2 | Chordee |
| | 2 | Chronic lung disease |
| | 2 | colpocephaly |
| | 2 | CONGESTIVE HEART FAILURE |
| | 2 | Conradi-Hunermann Syndrome |
| | 2 | cortical visual impairment |
| | 2 | Cri Du Chat |
| | 2 | Croup |

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| | 2 | Cushing's |
| | 2 | cyclopia |
| | 2 | dysgraphia |
| | 2 | Dysmotility of the intestines |
| | 2 | Eosinophilic Esophagitis |
| | 2 | epilepsy |
| | 2 | Giant Congenital Pigmented Nevus Leg |
| | 2 | glasses |
| | 2 | heart defect |
| | 2 | Hemimegalencephaly |
| | 2 | Holt-Oram |
| | 2 | Hyperpigmentation Of Gums |
| | 2 | hypoplastic aorta |
| | 2 | IGA deficiency |
| | 2 | infantile spasms |
| | 2 | infections |
| | 2 | inperforate anus |
| | 2 | interrupted aortic arch |
| | 2 | kyphosis |
| | 2 | L1CAM Syndrome |
| | 2 | Lamellar Ichthyosis |
| | 2 | large head |
| | 2 | latex precaution |
| | 2 | Ischnyhan |
| | 2 | lung |
| | 2 | lupus |

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|---|----------------------------------|
| 2 | Meckels Gruber |
| 2 | moles |
| 2 | mongolian spots |
| 2 | Neo Vaginal |
| 2 | Neutropenia |
| 2 | nevus |
| 2 | No Thyroid gland |
| 2 | patent Foramen Ovale |
| 2 | Peutz Jeghers Syndrome |
| 2 | PHPV |
| 2 | Pierre Robin Syndrome |
| 2 | Pilonidal Cyst |
| 2 | plastic tape |
| 2 | Polymicrogyria |
| 2 | Porencephaly |
| 2 | Precocious Puberty |
| 2 | Prune Belly |
| 2 | pulmonary sequestration |
| 2 | Russell-Silver |
| 2 | sensitive skin |
| 2 | Severe Combined Immunodeficiency |
| 2 | spinal cord |
| 2 | splenic cyst |
| 2 | split spinal cord |
| 2 | spinal fluid leak |
| 2 | STENOSIS |

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| 2 | stork bite |
| 2 | Sturge-Webber |
| 2 | Supraventricular Tachycardia |
| 2 | tape allergy |
| 2 | Tar |
| 2 | Taussig-Bing anomaly |
| 2 | tibial torsion |
| 2 | Tourettes' |
| 2 | tracheomalacia |
| 2 | Treacher Collins |
| 2 | two vessel cord |
| 2 | ulcerative colitis |
| 2 | ventriculomegaly |
| 2 | Vesicostomy |
| 2 | wide set nipples |

Reports by Category

| Group_Name | CountOfBirth_Defect_ID |
|------------------------|------------------------|
| Learning Disabilities | 1637 |
| Developmental Delay | 1265 |
| Allergies | 1232 |
| Gastrointestinal | 1176 |
| Cranial/Facial | 1104 |
| Central Nervous System | 1044 |
| Cardiovascular | 951 |
| Limb Defects | 890 |

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|--------------------------------------|-----|
| Immune Response Disorders | 890 |
| Emotional/Behavioral | 810 |
| Eye | 661 |
| Ear | 647 |
| Skin | 641 |
| Genitourinary | 561 |
| Syndromes | 517 |
| Spine | 421 |
| Muscle | 416 |
| Attention Deficit/Activity Disorders | 411 |
| Chest | 317 |
| Endocrine | 230 |
| Liver/Bile Duct/Spleen | 228 |
| Growth Disorders | 210 |
| Cysts | 113 |
| Tumors-Benign | 112 |
| Cancer | 24 |

Questionnaires Submitted by State

| CountOfReporter_ID | State |
|--------------------|-------|
| 140 | FL |
| 117 | TX |
| 112 | CA |
| 97 | OH |
| 76 | NY |
| 75 | IL |
| 63 | PA |
| 61 | MI |
| 56 | NC |
| 52 | TN |

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|----|-----------------|
| 50 | IN |
| 47 | GA |
| 47 | VA |
| 43 | NJ |
| 40 | MO |
| 38 | WA |
| 35 | MA |
| 35 | MN |
| 33 | KY |
| 32 | OR |
| 31 | AL |
| 31 | AR |
| 31 | MD |
| 31 | WI |
| 28 | IA |
| 26 | AZ |
| 25 | LA |
| 24 | OK |
| 22 | WV |
| 21 | CO |
| 20 | SC |
| 17 | KS |
| 15 | ID |
| 13 | NE |
| 12 | ME |
| 12 | NH |
| 12 | SD |
| 11 | CT |
| 10 | NV |
| 9 | UT |
| 8 | MT |
| 8 | NM |
| 8 | RI |
| 7 | DE |
| 7 | HI |
| 7 | MS |
| 3 | ND |
| 2 | AK |
| 2 | VT |
| 2 | WY |
| 79 | Other Countries |

64 | State Not Reported

3 | Guam

2 | Puerto Rico

1 | APO

1853