

# Autism Spectrum Disorders Report

From the National Birth Defect Registry

2026

## **Summary**

This report provides data from 390 Autism Spectrum Disorders cases in a database of 6727 cases of birth defects in the National Birth Defect Registry. The report also provides data on the high frequency of physical defects among children with Autism Spectrum Disorders. In addition, it includes most frequent parental exposures and illnesses recorded for pre-conception and pregnancy.

## **Background:**

The National Birth Defect Registry was started in 1990 by Birth Defect Research for Children (BDRC) to collect information on all kinds of birth defects both structural and functional, as well as information on the pre-conceptual and prenatal health, genetic factors and exposure histories of both parents. The Registry 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 BDRC. Data from the questionnaire were then manually entered into an inter-relational 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 made it easier to add new sections to the registry questionnaire without the cost of reprinting thousands of questionnaire booklets and mailing them to parents. The quality of data collection 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.

The registry is designed to identify patterns of similar birth defects that may have similar conditions or exposures in common in the parents' pregnancy or pre-conceptual history. A slide presentation [https://birthdefects.org/wp-content/uploads/2014/02/niehs\\_pres.pdf](https://birthdefects.org/wp-content/uploads/2014/02/niehs_pres.pdf) made to the National Institute of Environmental Health Sciences explains the operation of the registry and some of the birth defect issues BDRC has worked on using registry data.

Since April of 2004, parents have completed case reports for 6727 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 3452 males, 3254 females and 21 cases where the gender was not entered.

## **Report on Autism Spectrum Disorders in the National Birth Defect Registry**

Autism is a developmental disability that significantly affects verbal and nonverbal communication and social interaction. The symptoms of autism generally become evident before age three. ASDs are serious disorders of behavior and communication. In the mildest forms, ASDs may resemble learning disabilities. The more severe forms of autism involve self-injury and the need for constant care. Symptoms of autism are most often noticed in the first three years of life. Babies who resist cuddling and don't respond to their environment may be showing early signs of autism. To learn more about Autism Spectrum disorders, you can read our full fact sheet here:

<https://birthdefects.org/autism/>

ASDs have been increasing in incidence. Studies in late 1960's reported rates of ASD at 2-6 children per 10,000. According to the CDC's most recent estimate, 1 in 31 children aged 8 or younger will be diagnosed with ASD. The cause(s) of this increase are the subject of millions of dollars in new research in genetics and environmental exposures with no clear answers from either discipline.

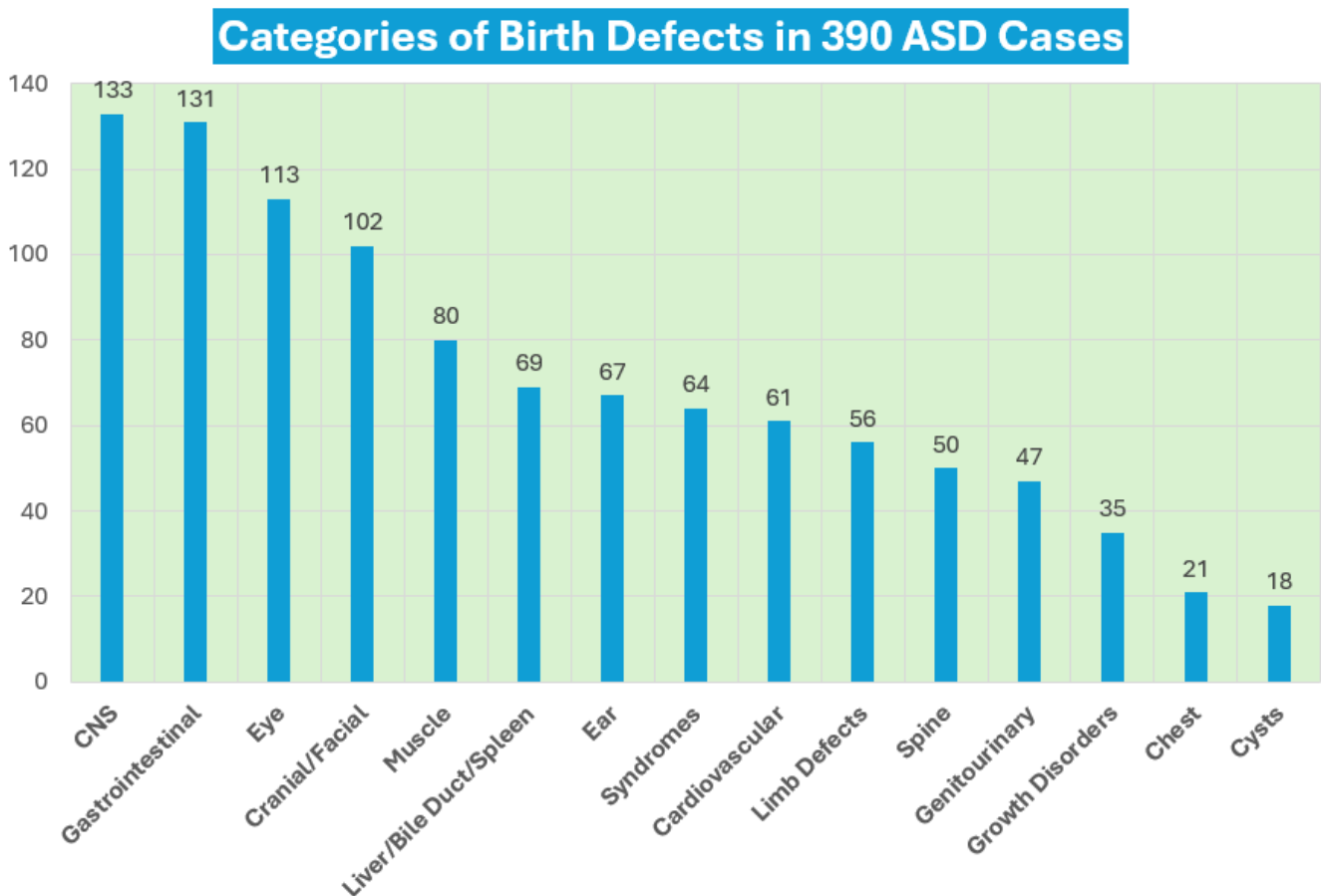
It is unclear whether ASDs are congenital defects; i.e., present at birth or acquired developmental problems that are associated with post-natal exposures.

### Most Frequent Birth Defects Reported by ASD Cases

Since the online version of the National Birth Defect Registry began, there have been 390 reports of Autism Spectrum Disorders. These cases were self-reported by the parents in the general public and through a special outreach to parents served by the Center for Autism and Related Disorders at the University of Central Florida (CARD). A case analysis has revealed that over 69% (269 cases\*) with Autistic Spectrum Disorders (ASDs) also had structural birth defects, primarily Central Nervous System (CNS) or Craniofacial Defects. 121 (31%) of the ASD cases reported associated developmental problems, but no structural birth defects.

Although continued case reporting could change the ratio of ASD/birth defect cases and ASD only cases, experience with Agent Orange and Gulf War data has shown that reporting patterns do not vary greatly with increased data collection.

**Chart 1: Birth Defects Reported in 390 ASDs in the National Birth Defect Registry**



<b>Related Muscle Defects</b>	
<b>Hypotonia (Floppy Muscles)</b>	57
<b>Muscle (Absent or Underdeveloped)</b>	22
<b>Hiatal Hernia</b>	8
<b>Fibromyalgia</b>	5
<b>Pectoralis (Absent or Underdeveloped)</b>	3
<b>Hypertonia</b>	2
<b>Diaphragmatic Hernia</b>	2
<b>Muscular Dystrophy</b>	2

<b>Related Central Nervous System Disabilities</b>	
<b>Seizure Disorder</b>	66
<b>Microcephaly</b>	19
<b>Cerebral Palsy</b>	18
<b>Hydrocephalus</b>	18
<b>Migraines</b>	18
<b>Chiari Malformation</b>	15
<b>Absence Corpus Callosum</b>	9
<b>Ataxia</b>	7
<b>Thin Corpus Callosum</b>	6
<b>Static Encephalopathy</b>	5
<b>Cortical Blindness</b>	5
<b>Dandy Walker Malformation</b>	4
<b>Periventricular Leukomalacia</b>	4
<b>Spina Bifida</b>	4
<b>Syringomyelia</b>	3
<b>Spastic Paralysis</b>	3
<b>Absence Of Septum Pellucidum</b>	3
<b>Spina Bifida Occulta</b>	3
<b>Neuropathy</b>	2
<b>Intrauterine Stroke</b>	2

<b>Related Cranial/Facial Disabilities</b>	
<b>Abnormal Teeth</b>	45
<b>High Arched Palate</b>	24
<b>Early Tooth Eruption</b>	24
<b>Swollen/Or Bleeding Gums</b>	19
<b>Facial Asymmetry</b>	15
<b>Micrognathia (Small Jaw)</b>	14
<b>Dysmorphic Features</b>	13
<b>Cleft Palate</b>	12
<b>Bony Defect of Skull</b>	11
<b>Craniosynostosis</b>	7
<b>Hypertelorism</b>	6
<b>Macrocephaly</b>	5
<b>Cleft Lip</b>	5
<b>Facial Bone Deformity</b>	5
<b>Facial Nerve Paralysis</b>	4
<b>Hyperpigmentation Of Gums</b>	3
<b>Temporomandibular Joint Disorder (TMJ)</b>	3

<b>Related Gastrointestinal Disabilities</b>	
<b>Reflux/GERD</b>	76
<b>Chronic Stomach Problems</b>	61
<b>Constipation/Chronic Constipation</b>	15
<b>Umbilical Hernia</b>	14
<b>Irritable Bowel Syndrome</b>	13
<b>Gastroschisis</b>	8
<b>Inguinal Hernia</b>	6
<b>Imperforate Anus</b>	5
<b>Pyloric Stenosis</b>	4
<b>Neurogenic Bowel</b>	3
<b>Tracheoesophageal Fistula/Esophageal Atresia</b>	3

<b>Related Eye Disabilities</b>	
<b>Vision Defect</b>	39
<b>Astigmatism</b>	36
<b>Strabismus</b>	30
<b>Amblyopia (Lazy Eye)</b>	22
<b>Optic Nerve Atrophy</b>	8
<b>Nystagmus</b>	8
<b>Blind</b>	7
<b>Exotropia</b>	7
<b>Cataract</b>	4
<b>Ptosis</b>	4
<b>Septo-Optic Dysplasia</b>	3
<b>Coloboma</b>	2

<b>Related Cardiovascular Disabilities</b>	
<b>Heart Murmur</b>	25
<b>Hypertension</b>	14
<b>Cardiac Arrhythmia</b>	11
<b>Atrial Septal Defect</b>	8
<b>Ventricular Septal Defect</b>	7
<b>Bradycardia</b>	4
<b>Pulmonary Valve Stenosis/Atresia</b>	4
<b>Aortic Valve Stenosis/Atresia</b>	3
<b>Mitral Valve Prolapse</b>	3
<b>Tetralogy Of Fallot</b>	3
<b>Pulmonary Hypertension</b>	3
<b>Bicuspid Aortic Valve</b>	2
<b>Hypoplastic Left Heart Syndrome</b>	2
<b>Transposition Great Arteries</b>	2
<b>Pulmonary Artery Anomaly</b>	2
<b>Vascular Ring</b>	2
<b>Patent Ductus Arteriosus</b>	2

<b>Related Syndromes</b>	
<b>Chromosome Disorder</b>	27
<b>Tourettes'</b>	7
<b>Goldenhar (Hemifacial Microsomia)</b>	4
<b>Fragile X</b>	4
<b>Ehlers-Danlos</b>	4
<b>Smith-Lemli-Opitz</b>	3
<b>Rett</b>	3
<b>Down</b>	3
<b>Trisomy</b>	2
<b>Lennox-Gastaut</b>	2
<b>CMV Virus</b>	2

<b>Related Ear Disabilities</b>	
<b>Partial Hearing Loss</b>	36
<b>Low Set Ears</b>	18
<b>Chronic Tinnitus (Ringing in Ears)</b>	13
<b>External Ear or Ears (Absent or Malformed)</b>	11
<b>Sensorineural Hearing Loss</b>	10
<b>Ear Tag(S)</b>	5
<b>Internal Ear or Ears (Absent or Malformed)</b>	4

<b>Related Cysts</b>	
<b>Brain Cyst</b>	8
<b>Ovarian Cyst</b>	7
<b>Ganglion Cyst</b>	3

<b>Related Liver/Bile Duct/Spleen Disabilities</b>	
<b>Jaundice At Birth</b>	61
<b>Liver Function Disorder</b>	6
<b>Absent Gallbladder</b>	4
<b>Hepatomegaly (Enlarged Liver)</b>	4
<b>Splenomegaly (Enlarged Spleen)</b>	2

<b>Related Spine Disabilities</b>	
<b>Scoliosis</b>	35
<b>Malformations Of Vertebrae</b>	9
<b>Tethered Spinal Cord</b>	8
<b>Spondylosis</b>	4
<b>Kyphosis</b>	3
<b>Spinal Muscular Atrophy</b>	3
<b>Spinal Stenosis</b>	3
<b>Torticollis</b>	3

<b>Related Genitourinary Disabilities</b>	
<b>Undescended Testicles</b>	20
<b>Small Sex Organs</b>	8
<b>Neurogenic Bladder</b>	8
<b>Malformed Sex Organs</b>	7
<b>Hypospadias</b>	6
<b>Malformed Kidney</b>	5
<b>Hydrocele</b>	5
<b>Absent Kidney (Renal Agenesis)</b>	4
<b>Malformed Ureter</b>	4

<b>Related Growth Disorders</b>	
<b>Constitutional Short Stature</b>	16
<b>In Utero Growth Retardation</b>	14
<b>Growth Hormone Deficiency</b>	5
<b>Failure to Thrive</b>	3

<b>Related Muscle Defects</b>	
<b>Hypotonia (Floppy Muscles)</b>	57
<b>Muscle (Absent or Underdeveloped)</b>	22
<b>Hiatal Hernia</b>	8
<b>Fibromyalgia</b>	5
<b>Pectoralis (Absent or Underdeveloped)</b>	3
<b>Hypertonia</b>	2
<b>Diaphragmatic Hernia</b>	2
<b>Muscular Dystrophy</b>	2

<b>Related Limb Defects</b>	
<b>Dislocated Hip/Hip Dysplasia</b>	13
<b>Malformed Fingers, Hands or Arms</b>	11
<b>Clubfoot</b>	8
<b>Malformed Toes, Feet or Legs</b>	6
<b>Twisted Lower Limb</b>	6
<b>Syndactyly (Webbed Fingers or Toes)</b>	6
<b>Contractures Of Elbows/Knees/Joints</b>	5
<b>Clubfoot With Bone Deformity</b>	5
<b>Overgrowth Of Upper Limb</b>	3
<b>Clinodactyly</b>	3
<b>Overgrowth Of Lower Limb</b>	2
<b>Reduction Deformity Lower Limb</b>	2
<b>Reduction Deformity Upper Limb</b>	2
<b>Clubbed Hands</b>	1
<b>Polydactyly (Extra Digits)</b>	1
<b>Chondromalacia Patella</b>	1
<b>Hemihypertrophy</b>	1

<b>Related Skin Disabilities</b>	
<b>Eczema</b>	81
<b>Acne-Like Rash</b>	39
<b>Strawberry Birth Mark</b>	19
<b>Vitiligo</b>	16
<b>Skin Fungus</b>	13
<b>Cafe Au Lait Birth Mark</b>	13
<b>Psoriasis</b>	10
<b>Changes In Skin Color</b>	7
<b>Port Wine Stain</b>	4
<b>Other Birthmark</b>	2
<b>Lamellar Ichthyosis</b>	1

**Chart 8: Demographic Statistics**

<b>Demographics</b>	
<b>Total Children with ASD</b>	390
<b>Male</b>	277
<b>Female</b>	112
<b>Not disclosed</b>	1
<b>Vietnam Veteran Father</b>	88
<b>Vietnam Veteran Mother</b>	2
<b>Gulf Veteran Father</b>	28
<b>Gulf Veteran Mother</b>	5

## Maternal Occupations During Pregnancy

The maternal workplace may be an important source of exposure to both illnesses and toxins. The medical and teaching professions place the prospective mother in contact with frequent illnesses and toxic exposures. Many studies of medical professionals have reported increases in birth defects and pregnancy loss. In the school setting, teachers and administrative workers have been exposed to pesticides, roof tarring and/or asbestos in many facilities. Restaurant workers may be exposed to pesticides because of health department requirements to control insects in cooking and food storage areas.

**Chart 10. Maternal Occupations**

<b>Mother Occupation During Pregnancy</b>	
<b>Homemaker/Did not work</b>	122
<b>Other/Not Disclosed</b>	39
<b>Administrative/Clerical</b>	36
<b>Education - K-12/College</b>	17
<b>Healthcare - PA/Nursing</b>	17
<b>Customer Service</b>	11
<b>Retail/Wholesale</b>	10
<b>Student</b>	10
<b>Information Technology</b>	9
<b>Hospitality - Restaurant/Bar</b>	8
<b>Daycare/Childcare/Caregiver</b>	7
<b>Sales</b>	7
<b>Manufacturing/Factory</b>	6
<b>Real Estate</b>	5
<b>Financial Services</b>	4
<b>Healthcare - Administrative</b>	4
<b>Legal</b>	4
<b>Military</b>	4
<b>Retail</b>	4
<b>Bank Teller</b>	3
<b>Manager</b>	3
<b>Automotive/Mechanic</b>	2
<b>Aviation Mechanic/Technician</b>	2
<b>Cosmetology (Hair/Nail/Beauty)</b>	2
<b>Engineering</b>	2
<b>Laboratory</b>	2
<b>Librarian</b>	2
<b>Operations Management</b>	2
<b>Pharmaceutical Sales</b>	2
<b>Public Relations</b>	2
<b>Respiratory Therapist</b>	2

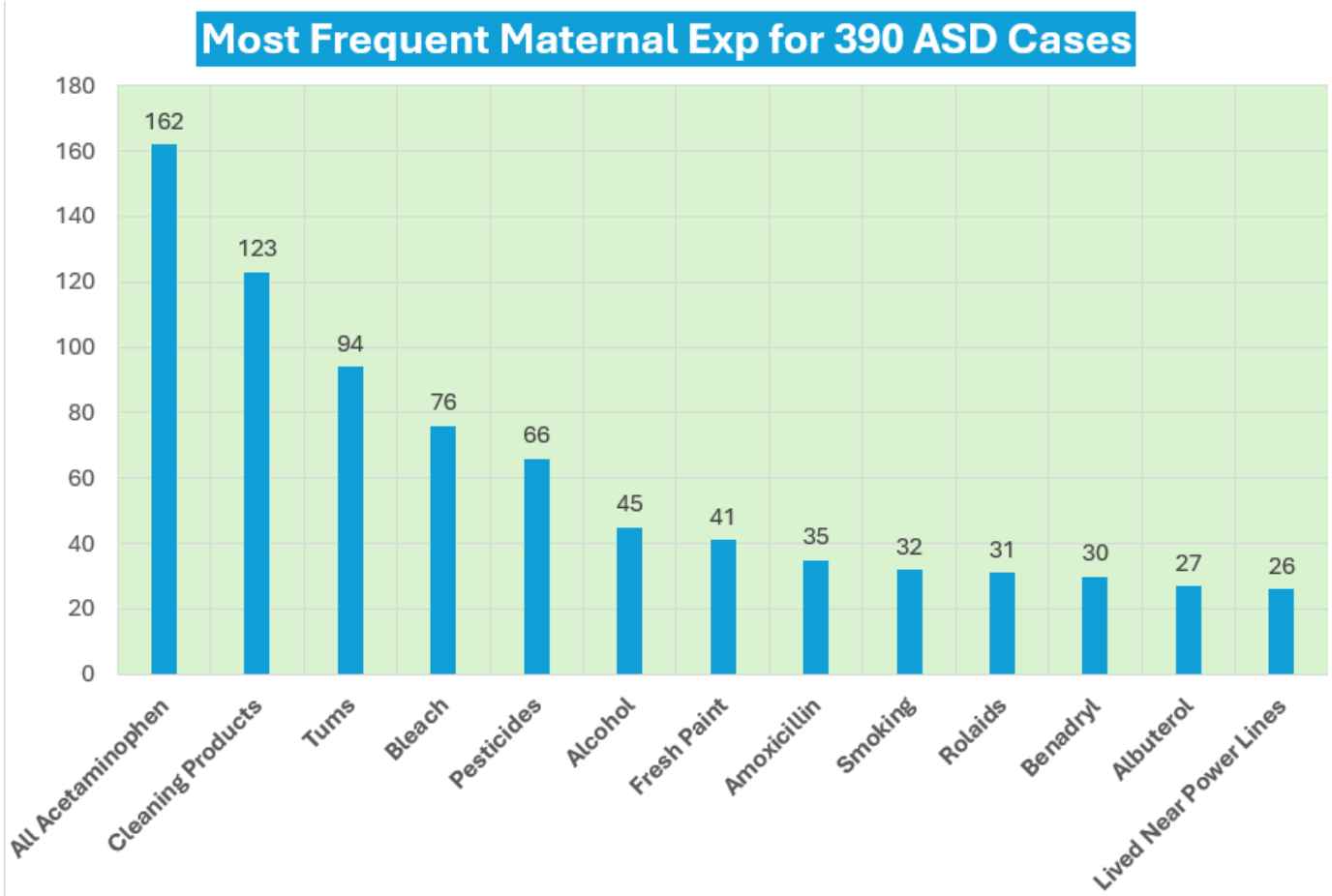
## Maternal Illnesses During Pregnancy

Maternal illness during pregnancy may expose the developing baby to viruses, bacterial infections, metabolic problems like diabetes and thyroid disorders and/or medications, X-rays and procedures used to treat maternal illnesses.

**Chart 11: Most Frequent Maternal Illnesses**

<b>Maternal Illness During Pregnancy</b>	
<b>Pregnancy Morning Sickness</b>	139
<b>Allergies</b>	84
<b>Headaches</b>	82
<b>Cold</b>	74
<b>Sinus Infection</b>	58
<b>Yeast Infection</b>	40
<b>Migraines</b>	40
<b>Sore Throat</b>	39
<b>Anemia</b>	28
<b>Urinary Tract Infection</b>	28
<b>Bronchitis</b>	25
<b>Flu</b>	22
<b>Gestational Diabetes</b>	21
<b>Preeclampsia</b>	19
<b>Virus</b>	11
<b>Vaginal/Pelvic Infections</b>	9
<b>Genital Herpes</b>	9
<b>Toxemia</b>	8
<b>Strep B</b>	8
<b>Strep Throat</b>	5
<b>Polyhydramnios</b>	4
<b>Kidney Infection</b>	4
<b>Chlamydia</b>	3
<b>Epstein Barr Virus</b>	3
<b>CMV</b>	2
<b>Pneumonia</b>	2
<b>Chronic Fatigue Syndrome (ME/CFS)</b>	2
<b>Rubella</b>	2

Chart 12: Most Frequent Maternal Exposures



Heavy Metals	
Mercury	4
Lead	4
Cadmium	1

Electromagnetic Fields	
Lived Near Power Lines	26
Lived Near Airport	14
Freq Electric Blanket	4

<b>Pesticides</b>	
<b>Prof Home/Office Spray Serv</b>	9
<b>Lived Near Lake/Power Lines/Canal</b>	9
<b>Lived within 3 miles of Agricultural Area</b>	8
<b>Raid</b>	8
<b>Roundup</b>	6
<b>Home Treated Termites/Ants</b>	4
<b>Applied Pesticides</b>	4
<b>DEET</b>	3
<b>Prof Lawn Service</b>	3
<b>Sevin</b>	3
<b>Malathion</b>	2

<b>Recreational Drugs</b>	
<b>Marijuana</b>	9
<b>Stimulants</b>	3
<b>Crack</b>	2
<b>Cocaine</b>	2
<b>Hallucinogen</b>	1

<b>Alcohol</b>	
<b>Occasional</b>	44
<b>Heavy Use</b>	1

<b>Smoking</b>	
<b>1-2 packs/day</b>	32
<b>Less than 1 pack/day</b>	29
<b>Second Hand Smoke</b>	3
<b>3+ packs/day</b>	1

<b>Worked At Computer</b>	
<b>Worked At Computer</b>	140

<b>Artificial Sweeteners</b>	
<b>Aspartame</b>	49
<b>Saccharin</b>	12
<b>Sucralose</b>	1

<b>Breast Implants</b>	
<b>Saline Breast Implants</b>	1
<b>Silicone Breast Implants</b>	1

**Mother Exposures: OTC and Prescription Drugs**

<b>Antifungals</b>	
<b>Fluconazole</b>	1
<b>Clotrimazole</b>	1

<b>Allergy Shots</b>	
<b>Allergy Shots</b>	7

<b>Anticonvulsants</b>	
<b>Depakote</b>	5
<b>Tegretol</b>	3
<b>Phenobarbital</b>	2

<b>All Acetaminophen</b>	
<b>All acetaminophen-containing medications combined</b>	162

<b>Allergy/Cold/Cough Medications</b>	
Benadryl	30
Sudafed	24
Robitussin	15
Nyquil	10
Tylenol Sinus	9
Claritin	9
Zyrtec	7
Dimetapp	4
Dristan	4
Actifed	2
Coricidin	2
Contac	2
Sinutab	1
Entex	1
Seldane	1
Alka Seltzer Gold	1
Chlortrimeton	1

<b>Heart Medications</b>	
Procardia	3

<b>Tranquilizers/Sedatives</b>	
Valium	2
Ambien	1
Xanax	1

<b>Blood Pressure Medications</b>	
Aldomet	2
Lopressor	1
Hydralazine	1
Lasix	1

<b>Nasal Sprays</b>	
Nasonex	7
Flonase	7
Afrin	5
Neosynephrine	3
Sinex	1

<b>Steroids</b>	
Prednisone	10

<b>Asthma Medications</b>	
Albuterol	27
Terbutaline	5
Proventil	2
Theophylline	1
Azmacort	1
Beclovent	1

<b>Antacids</b>	
Tums	94
Rolaids	31
Mylanta	23
Pepto Bismol	13
Gaviscon	7
Riopan	2
Digel	2

<b>Hormones</b>	
Progesterone	12
Oral Contraceptives	8

<b>Acne Medication</b>	
Accutane	1

<b>Antidepressants</b>	
Zoloft	11
Paxil	7
Wellbutrin	6
Prozac	6
Cymbalta	1
Sinequan	1

<b>Fertility Drugs</b>	
Clomid	7
Pergonal	1

<b>Antibiotics</b>	
Amoxicillin	35
Zithromax	13
Bactrim	6
Penicillin	5
Macrobid	5
Erythromycin	5
Keflex	4
Flagyl	3
Augmentin	3
Biaxin	2
Tetracycline	1
Ampicillin	1
Noroxin	1
Septra	1
Macrochantin	1

<b>Vaccines</b>	
Flu Shots	19
Tetanus, Diphtheria, Pertussis	3

<b>Diabetes Medication</b>	
Insulin	13
Diabeta	5

<b>Antipsychotics</b>	
Haldol	1

<b>Thyroid Medications</b>	
Synthroid	10
Levothyroid	3
Proloid	1

<b>Vaginal Medications/Contraceptives</b>	
Monistat	10
Nystatin	5
Chlotrimazole Cream	4
Spermicides	4
IUDs	3
Terazol	2
Gyne-Lotrimin	1
Betadine	1

<b>Pain Killers</b>	
Acetaminophen	149
Ibuprofen	22
Aspirin	13
Vicodin	9
Codeine	9
Demerol	7
Aleve	4
Morphine	2
Hydrocodone	2
Darvocet	2
Percodan	2
Naprosyn	2
Fentanyl	1

<b>Blood Thinners</b>	
Heparin	2

<b>Anti-nauseants/motion sickness medications</b>	
Zofran	18
Phenergan	18
Emetrol	5
Dramamine	4
Compazine	4
Bendectin	4
Meclizine	3
Scopolamine	2

<b>Anesthetics</b>	
Epidural/Spinal Block	8
General Anesthetic	4
Other Anesthetics	3
Local Anesthetic	1

<b>Asthma Medications</b>	
Albuterol	27
Terbutaline	5
Proventil	2
Theophylline	1
Azmacort	1
Beclovent	1

<b>Laxatives</b>	
Milk Of Magnesia	6
Metamucil	6
Dulcolax	6
Senokot	4
Correctol	1

<b>Chemical Exposure</b>	
<b>Cleaning Products</b>	123
<b>Bleach</b>	76
<b>Fresh Paint</b>	41
<b>Beauty Parlor Chem</b>	23
<b>Artificial Nail Tech</b>	15
<b>Diesel Fumes</b>	13
<b>Printing Ink</b>	10
<b>Commercial Art Supplies</b>	6
<b>Glues</b>	6
<b>Solvents</b>	5
<b>Asbestos</b>	4
<b>Mold/Black Mold/Mildew</b>	3
<b>Formaldehyde</b>	3
<b>Benzene</b>	2
<b>Toluene</b>	2
<b>Wood Stripper</b>	2
<b>Fiberglass Resin</b>	2
<b>Xylene</b>	2
<b>Tar</b>	1
<b>Photo Processing Chem</b>	1
<b>Trichlorethylene</b>	1

**Discussion:**

Sixty-nine percent of the ASD cases in the National Birth Defect Registry are associated with structural birth defects. The analysis of 390 cases may not reflect what would be found in a larger case collection, but if this ratio remains the same with continued reporting, registry data could suggest several possibilities. 1) ASDs are part of a birth defect pattern and those cases without reports of associated birth defects should be evaluated for subtle physical anomalies; or 2) There are two types of ASDs....one that occurs as part of a birth defect sequence and the other that may have an origin that may be either pre or post-natal; or 3) ASDs without associated birth defects may be the result of an expanded definition of autism that includes children with severe ADHD, Sensory Integration Disorders and other neuro-developmental problems that were not previously classified as autism. There might be a selection bias for structural defects in the people who choose to fill out the registry on our site, we would need to investigate this further to confirm these findings.

The pattern of illnesses reported in the Pre-conceptual year for both the mothers and the fathers of ASD cases in the registry is of interest since it could suggest a common disease process or exposures to similar environmental toxins.

The most commonly reported maternal exposures in the ASD cases were acetaminophen. More than two dozen studies have found association with Acetaminophen and ASDs and other neurodevelopmental disorders. This has resulted in actions from the FDA.

FDA Action (September 2025): The FDA began the process to update the product label for acetaminophen to reflect studies suggesting a potential association between, not a confirmed cause of, prenatal use and neurodevelopmental disorders like autism and ADHD. They also sent a "Dear Doctor" Letter/Notice to physicians to alert them to this emerging body of evidence.

## **Definitions Birth Defects**

**Ab Corpus Callosum** – absence of corpus callosum which is the band of tissue connecting the two hemispheres of the brain

**Abs septum pellucidum** – absence of the septum pellucidum, a thin membrane located at the midline of the brain.

**Anomalous Pulmonary Veins** - a rare congenital malformation in which all four pulmonary veins do not connect normally to the left atrium but instead drain abnormally to the right atrium by way of an abnormal (anomalous) connection.

**Anophthalmia** – absence of the eye or eyes

**Atrial Septal Defect (ASD)** – defect or opening between the heart's two upper chambers (the atria)

**BOR Syndrome** -Branchiootorenal (BOR) syndrome is a genetic condition that typically disrupts the development of tissues in the neck and causes malformations of the ears and kidneys.

**Bradycardia** - heart beats at an abnormally slow rate

**Clinodactyly** - a bending or curvature deformity of the finger which occurs in the plane of the hand typically caused by abnormal growth and development of the small bones of the finger (the phalanges)

**Coloboma** - a gap in part of the structures of the eye

**Cortical Blindness**-the total or partial loss of vision in a normal-appearing eye caused by damage to the visual area in the brain's occipital cortex

**Craniosynostosis** - a birth defect of the brain characterized by the premature closure of one or more of the fibrous joints between the bones of the skull (called the cranial sutures) before brain growth is complete

**Dandy Walker**- a congenital brain malformation involving the cerebellum (an area at the back of the brain that controls movement) and the fluid-filled spaces around it

**Diaphragmatic hernia**- an abnormal opening in the diaphragm, the muscle that helps you breathe. The opening allows part of the abdominal organs to move into the chest area.

**Goldenhar Syndrome**- abnormalities of the head and the bones of the spinal column. The abnormalities of the head can include differences with the eyes, ears, facial bones, and mouth and are usually more severe on one side.

**Hydrocephaly**-excessive accumulation of fluid in the brain

**Hydronephrosis** -distention (dilation) of the kidney with urine, caused by backward pressure on the kidney when the flow of urine is obstructed.

**Hypospadias**- a birth defect found in boys in which the urinary tract opening is not at the tip of the penis.

**Hemihypertrophy**- the enlargement of one side of the body or part of the body

**Imperforate Anus** - the opening to the *anus* is missing or blocked

**Kyphosis**- a forward curvature of the spine

**Lamellar Ichthyosis** – a rare inherited skin disorder

**Lissencephaly** -a rare, gene-linked brain malformation characterized by the absence of normal convolutions (folds) in the cerebral cortex and an abnormally small head (microcephaly).

**Macrocephaly**- a condition in which the head is larger than normal

**Microcephaly** – a condition in which the head is smaller than normal

**Micrognathia** - a condition where the jaw is undersized

**Organoid Nevus Syndrome**- a rare syndrome involving the skin, eye and brain.

**18 P Syndrome** - a frequent deletion *syndrome* characterized by dysmorphic features, growth deficiencies, and mental retardation

**Pectus Excavatum** – sunken chest wall

**Periventricular leukomalacia** – a white matter lesion in the brains of premature infants

**Pyloric stenosis** - a narrowing of the *pylorus*, the lower part of the stomach

**Septo-Optic-Dysplasia**- a syndrome characterized by absence of the midline part of the brain and underdevelopment of the optic nerve

**Severe combined immune deficiency**- a life-threatening syndrome of recurrent infections, diarrhea, dermatitis, and failure to thrive

**Spina Bifida** - a birth defect that involves the incomplete development of the spinal cord or its coverings

**Rett Syndrome** - a neurological and developmental disorder that mostly occurs in females

**Umbilical hernia** - an outward bulging (protrusion) of the abdominal lining or part of the abdominal organ(s) through the area around the belly button

**Undescended testicles** – a baby boy's testicle(s) have not moved into their proper position in the first few months of life

**Ureter Defect** – malformation of the tube(s) that connect the bladder to the kidney

**Ventricular Septal Defect** - A defect or opening between the heart's two lower chambers (the ventricles)

**XYY Syndrome** - an extra copy of the Y chromosome in each of a male's cells