

# NTD NEWSLETTER

January 2019  
Number 33



## NEURAL TUBE DEFECTS WITH OTHER ANOMALIES POSE GREATEST BARRIER TO COMPLETE PREVENTION

Among the 1,301 NTDs identified in South Carolina since 1992, 996 (77%) were isolated and 305 (23%) were associated with other malformations. It is the latter group – NTDs with other anomalies – that folic acid supplementation and fortification may be ineffective in preventing.

Specific causes of NTDs can be found most commonly among NTDs that are accompanied with other malformations. Chromosome aberrations are the most common cause identified among these cases (table below). Twenty four percent (72/305) of NTDs with other anomalies had chromosome defects, the

most frequent being Trisomy 18. Other recognizable patterns of malformations such as amniotic band syndrome and OEIS complex, (omphalocele, exstrophy, imperforate anus, and spine abnormalities) for which specific environmental or genetic causes have not been established accounted for 53/305 (17%) of the cases. Very few single gene causes of NTDs have been identified—Meckel syndrome being one most common.

A syndrome could not be recognized in half (155/305) of the NTD cases with other malformations.

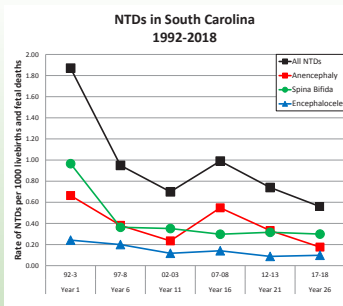
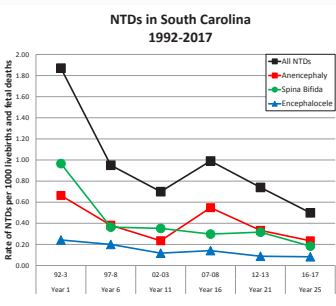
	Anencephaly and other anomalies	Spina Bifida and other anomalies	Encephalocele and other anomalies	All NTDs with other anomalies
All chromosomal aberrations	9	48	15	72
Trisomy 18	4	24	4	32
Trisomy 13		10	3	13
Trisomy 21		1		1
Other trisomy		2	2	4
Triploidy		7		7
Segmental deletion	1	1	3	5
Segmental duplication	1	1		2
Other	3	2	3	8
All single gene mutations	1	1	21	23
Meckel syndrome	1		18	19
Other		1	3	4
Sirenomelia	2	1		3
Amniotic bands	20	3	15	38
OEIS complex		10		10
Short Cord Complex/Limb-Body Wall Defect	1	1		2
Cause unknown/syndrome not recognized	58	65	32	155
TOTALS BY NTD TYPE	92	130	83	305

# Greenwood Genetic Center

101 Gregor Mendel Circle  
Greenwood, SC 29646

## GOVERNOR DECLARES JANUARY BIRTH DEFECTS AWARENESS MONTH

January 2019 has been designated by SC Governor Henry McMaster as Birth Defects Prevention Month. As part of Birth Defects Awareness Month, billboards have been placed throughout South Carolina in efforts to increase awareness about the importance of taking a multivitamin containing folic acid to prevent birth defects of the brain and spine. The message will be continued during the subsequent months through public lectures, science courses, press releases and exhibits.



Data from the past two calendar years are summarized below.

	2017	2018
Spina Bifida	13 (42%)	17 (53%)
Anencephaly	14 (45%)	10 (31%)
Encephalocele	4 (13%)	5 (16%)

Anencephaly was the most common NTD in 2017, spina bifida was the most common in 2018.

Ultrasound Scan	28	28
Quad Screening	1	2
Delivery	2	2

Most NTDs continue to be detected early in pregnancy by ultrasound.

Region 1 (Piedmont)	7	9
Region 2 (Midlands)	9	12
Region 3 (Coastal)	15	11

Distribution of cases by region during years 25 and 26.

### Contact Us:

To request free literature on NTDs and NTD prevention (English or Spanish) or to request a speaker at no charge for your office or group contact:

Jane H. Dean, RN

Statewide Program Coordinator

1-800-676-6332 or (864) 941-8138 or jane@ggc.org

Our dietary guideline with foods that are high in folic acid along with examples of menus using this information has proven to be a very helpful tool within the Hispanic communities.

### The Greenwood Genetic Center (GGC)

is a nonprofit institute organized to provide clinical genetic services and laboratory testing, to develop educational programs and materials, and to conduct research in the field of medical genetics. This newsletter is provided as an educational service to the health care community by GGC. For further information about the clinical and laboratory services available through the GGC please contact:

**Clinical Services:** 1-888-GGC-GENE  
(864) 941-8100 Greenwood  
(843) 588-4363 Charleston  
(803) 799-5390 Columbia  
(843) 664-2983 Florence  
(864) 250-7944 Greenville

**Fetal Examination** (864) 388-1700  
(864) 388-1707 (fax)

**Laboratories** 1-800-473-9411  
(864) 941-8110



### SC Birth Defects Prevention Program Partners

**Greenwood Genetic Center** - Jane Dean, RN (Coordinator)  
(864) 941-8138, 1-800-676-6332

**Shriners Hospitals for Children** (Greenville) - Myelodysplasia Services  
(864) 271-3444

**SC Department of Disabilities and Special Needs**  
(803) 898-9789

**SC DHEC - SC Birth Defects Surveillance Program** - Vinita Oberoi Leedom  
(803) 898-0771

Contributions to support the Birth Defects Prevention Program may be made through the Greenwood Genetic Center Foundation ([www.ggc.org/foundation](http://www.ggc.org/foundation) or 864-388-1813)