



SOX3 Duplications and Lumbosacral Spina Bifida

Chromosomal aneuploidies, microduplications and microdeletions are the most common confirmed genetic causes of spina bifida. Microduplications of Xq27 containing the SOX3 gene have been reported in eleven cases, confirming the existence of an X-chromosome locus for spina bifida. A three generation family with a SOX3 duplication has been identified in one of the seventeen families with recurrences in the 29 years of the South Carolina Neural Tube Defect Prevention Program. This family further confirms the association of spina bifida and duplication of the X chromosomal gene SOX3. Other recurrences during this time period included siblings with an *APAF1* mutation, siblings with a *CASP9* mutation, siblings with a microdeletion of 13q, and two sets of siblings with Meckel Syndrome.

The precise frequency of chromosome aberrations in isolated NTDs has not been established but is less than five percent. The frequency of cytogenetic/cytogenomic aberrations among 300 NTDs with other malformations was reported by Dean, Pauly and Stevenson (2020) to be 25.7% (77/300).

Request for NTD-Affected Tissues

Recent research has found that the gene alterations responsible for certain birth defects are only found in affected tissues. In order to determine if this is true for NTDs, we request referral of NTD-affected fetuses or stillbirths to the Greenwood Genetic Center Fetal Examination Service to collect the appropriate tissues for gene sequencing and microarray analysis. There are no costs to the family for this examination and testing.

Contacts for the Fetal Examination Service:

<i>Jennifer Claphan</i>	<i>Anna Crocket</i>
<i>Fetal Exam Technician</i>	<i>Administrative Assistant</i>
<i>(864) 941-8148</i>	<i>(864) 941-8103</i>

January 2022 Declared Birth Defects Awareness Month

January 2022 has been designated Governor Henry McMaster as Birth Defects Awareness Month in South Carolina.

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.

Recommendation for Increasing Folic Acid

The Neural Tube Defect Prevention Program at the Greenwood Genetic Center has proposed that ***the standard preventive dose of folic acid for all women of childbearing age in South Carolina be increased to 4 mg/day during the 3 months prior to conception and the first month after conception.***

South Carolina is a high-risk state for these defects, and 4 mg/day is currently the dose recommended by the CDC for high risk pregnancies (those with a prior affected infant).

This increased dose should be used with two caveats.

(1) The folic acid should be prescribed as **folic acid alone** rather than by increasing the number of multivitamins, as the dose of other vitamins may become excessive. This increased dose of folic acid may be used in addition to a single multivitamin with 0.4 mg folic acid per day.

(2) If any side effects should occur, discontinue the 4 mg/day dose and return to the multivitamin with only 0.4 mg/day or prenatal vitamin with 0.8 mg/day alone.

Greenwood Genetic Center

101 Gregor Mendel Circle
Greenwood, SC 29646

Thinking about having
a baby?

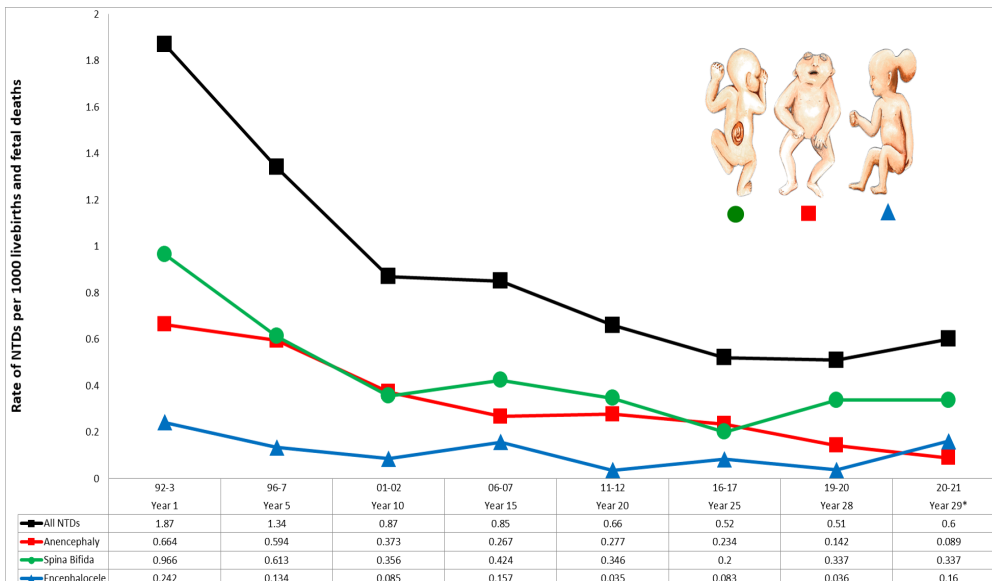


SC Department of Disabilities and Special Needs • SC Department of Health and Environmental Control • Greenwood Genetic Center

You need more
FOLIC ACID
than you think!

Call your doctor today or contact the
SC Birth Defects Prevention Program
1-800-6-SOMEDAY

Neural Tube Defects in South Carolina (1992-2021)



NTDs in SC during 2021 (Project year 29)

Cases by type of NTD:

Spina bifida	20 (59%)
Anencephaly	5 (15%)
Encephalocele	9 (27%)

Detection method:

Ultrasound	28
Quad screen	2
Fetal MRI	2
Delivery	2

Geographic Distribution

Region I (Piedmont)	12
Region II (Midlands)	7
Region III (Coastal)	15

*Year 29 data is preliminary pending final review

Contact Us:

Staff members at the office of the South Carolina Birth Defects Prevention Program will be happy to assist your office in any way to assure that your patients have information regarding prevention of these serious defects. We have literature on NTDs and NTD prevention available in both English and Spanish. A 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 with the Hispanic communities.

Jane H. Dean, RN

Statewide Program Coordinator

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

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