Neural Tube Defects Registry
First Multi-institutional Report 2006
King Faisal Specialist Hospital & Research Centre, Riyadh
Registration from (2000-2006)
and
Disable Children Association, Riyadh
Registration from (2006)

Executive Summary
The Neural Tube Defects Registry
The King Faisal Specialist Hospital and Research Centre (KFSH&RC) established a registry in March 2000 for all patients with neural tube defects presenting to the hospital. The registry is a coordinated collaboration among the departments of Neurosciences, Biostatistics, Epidemiology and Scientific Computing (BESC), Pediatrics, Orthopedics, Urology, and Obstetrics and Gynecology (Ob-Gyn). The development of the registry is meant to provide a better understanding on disease occurrence and natural history of the disease. Last year we started collaborating with the Disabled Children's Association in an effort to develop this registry nationally.
Data in this report is divided into two parts. The first covers data on the Disabled Children's Association and the second on King Faisal Specialist Hospital and Research Centre.

The Disabled Children's Association
A total of 39 patients were registered from March 1 to December 31, 2006 in the Neural Tube Defects registry of the Disabled Children's Association. The registry sample are all Saudi's with (n=21; 54%) females and (n=18; 46%) males. Most of the patients are attending school (n=10; 63%). Out of the 10 patients 8 are going to primary school. Despite the sonography most of the defects were reported to be first diagnosed at birth and during third trimester (n=15; 43%) and (n=12; 34%) respectively. The majority of the patients are diagnosed with **Spina Bifida** Aperta and Hydrocephalus (n=31; 80%). Among patients who were born with hydrocephalus (n=19; 66%) were diagnosed antenataly and (n=10; 35%) were diagnosed after birth. The majority (n=24; 67%) had operation of the primary defect on the first and second day of life. Among the 31 patients requiring VPS insertion (n=25; 89%) patients had the VP inserted after the repair.
None of the women in the registry sample took folic acid before conception and (n=25; 77%) of the mothers did not take any folic acid during the first three months of pregnancy. Around half the number of parents (n=19; 53%) stated they were not related, while (n=17; 47%) stated that they were related. Most of the patients (n=34; 94%) did not have a first degree relative with NTD. Moreover (n=27; 79%) patients did not have blood relatives with NTD.

The King Faisal Specialist Hospital and research centre
A total of 444 cases were registered from King Faisal Specialist Hospital and Research Centre from October 2000 to December 31, 2006. A total of (n=416; 94%) are Saudis with (n=202; 46%) males and (n=242; 55%) females. Twenty seven (n=27; 53%) patients are attending school, with (n=20; 39%) who did not reach school age. Around (n=20; 74%) of these patients are attending primary school. The majority of patients (n=50; 98%) are not employed and did not reach employment age. Most of the patients cannot move around because they are handicapped (n=37; 73%), and (n=11; 22%) of these patients move around independently in the house and community.

Among the mothers who had ultrasound, (n=306; 72%) of the mothers reported that their children were diagnosed with NTD at birth and (n=76; 18%) during the third trimester. Out of the 444 patients (n=271; 61%) had **spina bifida** aperta with hydrocephalus. With the relevant population being those women who had ultrasound, (n=109; 48%) were diagnosed with hydrocephalus before birth and (n=118; 52%) were diagnosed after birth. The majority of patients underwent the primary operation within the first three days of life (n=219; 53%). Around (n=268; 61%) had VPS inserted, while (n=174; 39%) did not have any. Among patients who had VPS inserted (n=195; 76%) had it after repair of the defect. The majority of our registry patients did not sustain any fractures (n=44; 88%).
A large number of mothers did not take folic preconception (n=407; 99%). The first trimester (n=348; 87%) had no folic acid intake as well, with (n=45; 11%) not taking this vitamin regularly. A good number of parents declared they were not related (n=243; 57%), while (n=180; 43%) declared they were**.** The vast majority of patients did not have first degree relatives with NTDs (n=418; 98%). Also a significant percentage of patients (n=403; 95%) did not have blood relatives with NTD.

Foreword

I am proud to release the Annual Report for 2006 for the Neural Tube Defect Registry. A total of 444 cases have been registered since this Registry was established in 2000, in collaboration with Biostatistics, Epidemiology and Scientific Computing, Pediatrics, Orthopedics, Urology and Obstetrics & Gynecology at the King Faisal Specialist Hospital & Research Centre.

The Registry is useful for identifying risk factors, diagnosis of NTDs and management of ***spina bifida****. The Neural Tube Defects Registry provides vital information in accumulating data that provides statistical and demographical information on prevalence, incidence and clinical data on NTDs in Saudi Arabia.*

Data collection provides an effective foundation to create national programs, health policies and prevention measures as well as promoting the welfare of infants and children, including outreach activities and patient education information. The Registry has also recently formed collaborations with local institutions in an effort to expand to a National Registry.

I would like to express my appreciation to the King Faisal Specialist Hospital & Research Centre for support and resource assistance on this project, as well as the Disabled Children's Association and Riyadh Central Hospital (Shemaisi) for promoting and supporting this Registry on a national level.

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Acknowledgements

We would like to deeply thank Dr. Essam Al Shail, Section Head, Neurosurgery and
Chairman of the Neural Tube defects Registry, also Dr. K. Ravichandran, Post Doctoral
Fellow, and Epidemiologist, for their assistance throughout the editing process of this report.
Special thanks to Dr. Zayyed Al Zayyed, Chairman Orthopedic Surgery Department and to
Dr. Mohammed Al Abdulaaly, Urology Consultant for providing resources of data collection.
We would like to extend our profound gratitude to Dr. Sultan Al Sedairy, Executive Director
of the Research Centre for his continued support in provision of resources.
We would also like to thank Ms. Shazia Subhani, Head, Registries Core Facility for her
continued support and professional guidance. Special acknowledgement to Ms. Samia Al
Hashim, for her contribution of technical expertise and assistance. Finally we gratefully
acknowledge Ms. Ihsan Yassen, NTDR Registrar, for her hard work and perseverance.

Registry Committee

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Neural tube defects including anencephaly, encephalocele and **spina bifida** are major birth defects of the brain and spine. These birth defects can cause lifelong disability or death. **Spina bifida** is the incomplete formation of the spine and the spinal cord. Its manifestations may include paralysis and loss of sensation of the legs. It also includes bladder and bowel incontinence. Moreover, it results in hydrocephalus which may affect intelligence, visual, hearing and learning disabilities.

Anencephaly involves absence of the skull bones, and part or

all of the brain. Anencephaly is always lethal. It usually results in either stillbirth or death within hours or days of birth. Encephalocele is a protrusion of the brain through a defect in the skull. These defects can occur early in pregnancy long before a woman realizes she is pregnant.

Each year **spina bifida** and anencephaly, the two most common forms of neural tube defects, occur in approximately 1 of every 1,000 pregnancies in the United States and in an estimated 300,000 newborns worldwide (CDC, 2007).

Risk Factors

Women are at greater risk of having a pregnancy affected by **spina bifida** or another neural tube defect (NTD) if they have:

A child with **spina bifida** or a previous pregnancy affected by any NTD. An NTD-affected pregnancy increases a woman's chance to have another NTD-affected pregnancy approximately twenty times

Maternal diabetes with uncontrolled blood glucose

Use of certain anti-seizure medication (e.g. Valproic acid/Depakene and Carbamazapine/Tegretol)

Medically diagnosed obesity (BMI >30)

Exposure to high temperatures in early pregnancy (i.e., prolonged fevers and hot tub use)

Race/ethnicity (Hispanics in the U.S are known to have the highest prevalence rate of NTD's)

Consanguinity

Mother's age (NTD risk is highest among youngest and oldest women)

Folic acid deficiency

Some of these risk factors are discussed in this report.

Diagnosis of NTD's and Ultasonography

Most NTD's are usually detected before a baby is born if a mother has an elevated alpha fetoprotein (AFP) level, a diagnostic blood test performed at about 16 weeks gestation, the amount of time after conception. This is then confirmed by an ultrasound which is used to create images of the fetus and the position of the spinal opening.

Spina bifida repair

Spina bifida repair is usually initiated within the first few days of life to reduce the risk of further damage to the spinal cord and nerves. Furthermore, most patients with spina bifida develop hydrocephalus, approximately 85-90%. Hydrocephalus is usually treated by surgically inserting a Ventriculoperitoneal shunt which redirects the fluid from the brain to the abdomen.

Folic acid

Any woman who can become pregnant is at risk of having an NTD affected pregnancy. Over the last two decades accumulating evidence has made it clear that the use of periconceptional of folic acid can significantly reduce the risk of NTD affected pregnancies (Enaw et al., 2006). Women who take folic acid everyday decrease their chance of having an affected child by 50-70%. Taking enough folic acid is not a guarantee; however other factors can play a role including environmental and genetic factors.

The future of spina bifida patients

Spina bifida is often complicated by physical and mental disabilities in different forms. Therefore, long term rehabilitation is very important to help patients establish independence in their daily activities (Suyama et al., 2000). Participation in school and community helps them acquire knowledge of dealing with social life and eventually promoting their social independence. Training for employment is an excellent factor that encourages independence and is very essential to help them take on responsibilities that are within their

capabilities.
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In March 2000, the King Faisal Specialist Hospital and Research Centre (KFSH&RC) established a registry for all patients with neural tube defects. The registry is a coordinated collaboration among the departments of Neurosciences, Biostatistics, Epidemiology and Scientific Computing (BESC), Pediatrics, Orthopedics, Urology, and Obstetrics and Gynecology (Ob-Gyn). The development of the registry is meant to provide a better understanding of disease occurrence and natural history of the disease. It serves as a base for epidemiological research and it is a start towards control and prevention. It includes all patients presenting to the hospital who are diagnosed with NTD. These comprise Saudi and non Saudi patients.

Data sources and collection

Data is usually collected in two phases. The first phase takes place in the clinic where patients' parents are interviewed face to face. The second phase involves further information about patients' diagnosis which is found in their medical charts. Data about diagnosis are usually coded using the British Pediatric Association's (BPA) classification of diseases, the World Health Organization's International Classification of Diseases 9th Revision and Clinical Modification (ICD-9-CM). The Combined **Spina Bifida** clinic is a

source of patient capture. This clinic is held twice a month. Approximately 7 patients in this clinic are seen by a team of specialty clinicians. Additionally, the registrar regularly visits the Urology and Pediatric clinics to capture all the NTD cases presenting to the hospital.

Data processing

Data collected is entered into database on MS SQL server 2000. Quarterly audits took place to assure legibility, completeness, and consistency of data items in all sections of the registry. The auditing function provides a mean to achieve verification of data from medical charts. Finally, data analysis is done using SAS and SPSS statistical packages.

Confidentiality

Information collected by the Registry is kept confidential using computer security measures and locked cabinets. All staff members are required to sign a pledge to maintain the confidentiality of all information collected. Confidentiality is strictly preserved so that the rights and welfare of the patients and families are not compromised.

Recent Achievements

Last year the registry has expanded its activities to include the Disabled Children's Association with which we started collaborating in an effort to expand the registry nationwide. We also signed a collaborative agreement with Riyadh Central Hospital (Shemaisi Hospital).

In June 2006, the NTDR committee took a decision to modify the baseline demographic component by adding 4 new variables. Data on these variables are collected retrospectively and on new patients. The variables are: education, employment, Mobility and

fractures.
All these variables are included under the demographic section, except for fractures, which is included under Clinical data.

The Disabled children's Association

Disabled Children's Association (DCA) is a non-profit non-government, charity organization working in Riyadh for many years with five more centers scattered all over the Kingdom (Makkah, Madina, Jouf, Jeddah and Hail) and two more under construction. The DCA registers around 20 to 30 newly diagnosed NTD patients per year. There are around 200 existing cases; most of them have been treated at King Faisal Specialist Hospital and Research Center (KFSH&RC).

Report overview

This report consists of two parts. The first belongs to Disabled Children's Association and the second belongs to King Faisal Hospital and Research Centre. Each part is divided into sections which are presented in tables and charts.
Sections include data presentation on patients' demographics, and Antenatal diagnosis of neural tube defects and hydrocephalus. In addition, data presentation includes clinical information and risk factors.

A total of 39 cases were registered from March 1 to December 31, 2006 in the Neural Tube Defects registry at the Disabled Children's Association. Five patients from outside of the Riyadh region on whom we have limited data, have been registered and included in this analysis. Data belonging to DCA is mainly presented in tables. Furthermore, a total of 444 were registered at the King

Faisal Specialist Hospital and Research Centre from October 2000 to December 31, 2006. Most of the data belonging to KFSH&RC in this report are presented in tables and charts. The new variables are presented only in tables due to the small number of cases that provided this information. We could retrieve data on these new variables for only 16 patients from DCA and 51 patients from KFSH&RC.

Part I: Disabled Children's Association

Section1.0: Patient Demographics

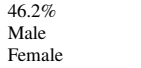
Gender distribution of NTD Saudi patients

Our registry sample in table 1.1 is all Saudi with 21 females (54%) and 18 males (46%).

Table 1.1: Gender distribution of Saudi patients

Gender	**Count**	**Valid percentage**
Male	18	46.2
Female	21	53.8
Total	**39**	**100.0**

Figure 1.1: Distribution of Saudi patients by gender



Regional distribution

As shown in Table 1.2 and 1.3, data is collected for both patrilocality and current residence with Riyadh topping both lists. Patrilocality documents the birth place of the father.

Table 1.2: Distribution of NTD patients by current domicile

Riyadh	35	94.6
Qasim	1	2.7
Jizan	1	2.7

Total Saudi Cities
37
100.0

Table 1.3: Distribution of NTD patients by patrilocality

Riyadh	20	55.6
Qasim	6	16.7
Jizan	3	8.3
Al Jouf	1	2.8
Northern Province	1	2.8
Baha	1	2.8
Makkah	1	2.8
Eastern Province	1	2.8
Tabuk	1	2.8
Madina	1	2.8
Total Saudi Cities	**36**	**100.0**

Education

Most of the patients are attending school 10 (63%), 5 patients are considered as not applicable the "not applicable" code was added to indicate patients who did not reach school age yet (Table 1.4). Out of the 10 patients 8 are going to primary school (Table 1.5).

Table 1.4: Distribution of NTD patients by education

Yes	10	62.5
Disabled	1	

6.3
Not Applicable
5
31.3
Total
16
100.0
Table 1.5: Distribution of NTD patients by education level

Pre-school
1
11.1
Primary
8
88.9
Total
9
100.0
Employment and Mobility
We did not display tables for **employment and mobility** because all the 16 patients did not reach employment age and are non walkers (100%).

Section 2.0: Antenatal Diagnosis of Neural Tube Defects and Hydrocephalus

Ultrasonography and stage of NTD diagnosis

Table 2.1 presents the number of mothers reporting ultrasonography by time of diagnosis. Despite the sonography, most of the defects were reported to be first diagnosed at birth and during third trimester 15(43%) and 12 (34%).

Table 2.1: Mothers with ultrasonography and stage of NTD diagnosis

Second trimester
8
22.9
Third trimester
12
34.3
At birth
15
42.9
Total
35
100.0

Diagnosis

In table 2.2 the majority of the patients are diagnosed with **Spina Bifida** Aperta and Hydrocephalus 31(80%).

Table 2.2: Distribution of patients by diagnosis

Spina bifida aperta, any site, with hydrocephalus	31	79.5
Spina bifida of any unspecified type with hydrocephalus	1	2.6
Spina bifida aperta, without hydrocephalus	7	17.9
Total	**39**	**100.0**

Hydrocephalus and antenatal diagnosis

Table 2.3 shows that 31(80%) patients are born with hydrocephalus; with 19(66%) diagnosed antenataly and 10(35%) diagnosed after birth (Table 2.4).

Table 2.3: Distribution of patients born with hydrocephalus

Yes	31	79.5
No	8	20.5
Total	**39**	**100.0**

Table 2.4: Distribution of patients with hydrocephalus by stage of 1st diagnosis

Diagnosed prior to birth	19	65.5
Diagnosed post birth	10	34.5
Total		

29
100.0
9

Section 3.0: Clinical Data History

Age of repair (in days)

Table 3.1 presents age in days at which the patients underwent the primary repair. The majority 24(67%) had their operation on the first and second day of life.

Table 3.1: Distribution of patients' age of repair

1 day
16
44.4
2 days
8
22.2
4-29 days
7
19.4
> 30 days
5
13.9
Total
36
100.0

Ventriculoperitoneal shunt

Table 3.2 shows the number of patients requiring a ventricular shunt apparatus 31(82%).

Table 3.2: Distribution of patients requiring VPS insertion

Yes
31
81.6
No
7
18.4
Total
38
100.0

Stage of VPS insertion

Among the 31 patients requiring VPS insertion 25 (89%) patients had the VP inserted after the repair. It is a standard procedure to have VPS inserted after the repair (Table 3.3).

Table 3.3: Distribution of patients by stage of VPS insertion

At the repair
1
3.6
After the repair
25
89.3
Before the repair
2
7.1
Total
28
100.0

Fractures

We did not show the table for fractures because we could only collect information for 14 patients, who all happened to be with no fractures (100%).

Section 4.0: Risk Factors

Folic acid intake

Looking at table 4.1 we find that none of the women in the registry sample took folic acid before conception. While 25(77%) of the mothers did not take any folic acid during the first three months of pregnancy, eight mothers took folic acid on and off.

Table 4.1: Distribution of women taking folic acid preconception and 1st trimester

Count
Valid percentage
Count
Valid percentage
On/Off
-
-
8
23.5
No
34
100.0
25
76.5
Total
34
100.0
34
100.0

Code On/Off was added to identify mothers who did not take folic acid regularly

Consanguinity

Nineteen parents stated they were not related 53%, while 17 stated that they were related 47% (Table 4.2).

Table 4.2: Consanguinity distribution of NTD patients

No consanguinity
19
52.8
Related
17
47.2
Total
36
100.0

First degree relative and blood relative with NTD

In table 4.3, we find that 34(94%) of the patients did not have a first degree relative with NTD. Moreover 27(79%) patients did not have blood relatives with NTD.

Table 4.3: Distribution of patients having first degree relative and blood relative with NTD

Count
Valid percentage
Count
Valid percentage
Yes
2
5.6
7
20.6
No
34
94.4
27
79.4
Total
36
100.0
34
100.0

First degree relative is defined as: Mother, Father, Sibling, Half-sibling or Child

Age of mothers

Table 4.4 presents age of mothers when the patients were born, with the highest number of mothers being between 21-30 years of age 21(54%).

Table 4.4: Distribution of age of mothers at birth of child

15-20
3
7.7
21-30
21
53.8
31-40
13
33.3
41-50
2
5.1
Total
39
100.0
11

Part II: (King Faisal Specialist Hospital & Research centre)

Section 1.0: Patient Demographics

Table 1.1: Distribution o f NTD patients by nationality

Saudi
416
93.7
Other Arab
26
5.9
Non Arab
2
0.5
Total
444
100.0

Figure 1.1: Distribution of NTD patients by nationality

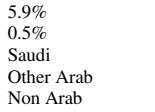
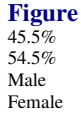


Table 1.2: Distribution o f NTD patients by gender

Male
202
45.5
Female
242
54.5
Total
444

100.0

Figure 1.2: Distribution o f NTD patients by gender



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Nationality and gender distribution

Table 1.1 and figure 1.1 shows the number of registered NTD patients by nationality, 94% are Saudis with 46% males
and 55% females (Table 1.2 and Figure 1.2).

Table 1.3: Distribution of NTD patients by current residence

Riyadh
174
40.2
Eastern Province
80
18.5
Makka
37
8.5
Madina
33
7.6
Qasim
17
3.9
Jizan
16
3.7
Tabuk
15
3.5
Al Jouf
14
3.2
Hail
13
3.0
Northern Province
12
2.8
Asir
10
2.3
Najran
7
1.6
Baha
5
1.2
Total Saudi Cities
433

98.2
Other Cities
8
1.8
Total
441
100.0

Table 1.4: Distribution of NTD patients by patrilocality

Riyadh
107
26.4
Eastern Province
59
14.5
Qasim
45
11.1
Makka
38
9.4
Madina
34
8.4
Hail
23
5.7
Asir
21
5.2
Jizan
19
4.7
Al Jouf
15
3.7
Northern Province
12
3.0
Najran
12
3.0
Tabuk
11
2.7
Baha
10
2.5
Total Saudi Cities
406
92.3
Other Cities
34
7.7
Total
440

100.0
13

Regional distribution

The data on current residence and patrilocality is shown in table 1.3 and 1.4. The geographical locations of NTD
patients are shown in descending order with Riyadh and Eastern Province ranking first on both lists.

Education

In table 1.5, 27(53%) patients are in school, with 20 (39%) who did not reach school age to whom the "not applicable" code applies. Around 20(74%) of these patients are attending primary school (Table 1.6).

Table 1.5: Distribution of NTD patients by education

Yes
27
52.9
No
1
2.0
Completed
1
2.0
Disabled
2
3.9
Not Applicable
20
39.1
Total
51
100.0

Table 1.6: Distribution of NTD patients by education level

Pre-school
2
7.4
Primary
20
74.1
Intermediate
3
11.1
Secondary
2
7.4
Total
27
100.0

Employment

The majority of patients are not employed. Most of

them are in school and did not reach employment age 50(98%), with only one patient who is not working (Table 1.7)

Table 1.7: Distribution of NTD patients by employment

No
1
2.0
Not Applicable
50
98.0
Total
51
100.0

Mobility

Most of the patients cannot move around because they are handicapped 37(73%), and 11(22%) of these patients move around independently in the house and community (Table 1.8). Only 3 patients did not reach walking age to which the "not applicable" code applies.

Table 1.8: Mobility

Non walker
37
72.5
Household walker
2
3.9
Community walker
9
17.6
Not applicable
3
5.9
Total
51
100.0
14

Section 2.0: Antenatal Diagnosis of Neural Tube Defects and Hydrocephalus

Ultrasonography and stage of NTD diagnosis

Among the mothers who had ultrasound, 72% of the mothers reported that their children were diagnosed at birth and 18% during the third trimester. Only 1% and 9% were diagnosed during the first and second trimester (Figure 2.1).

Figure 2.1: Mothers with ultrasonography and stage

of NTD diagnosis
72.0%
17.9%
9.4%
0.7%
First Trimester
Second Trimester
Third Trimester
At Birth

Diagnosis

Most of the cases presenting to KFSH&RC are Myelomeningoceles (i.e. **spina bifida** aperta). Out of the 444 patients (61%) had **spina bifida** aperta with hydrocephalus. As for **spina bifida** aperta without hydrocephalus we have 97(22%) cases, 48(11%) patients with lipomyelomeningocele and 9(2%) with occipital encephalocele (Table 2.2).

Table 2.2: Distribution of patients by diagnosis

Anencephaly
1
0.2
Spina Bifida Aperta with Hydrocephalus
271
61.0
Spina Bifida Aperta with Hydrocephalus, Parietal Encephalocele
1
0.2
Spina Bifida Aperta with Hydrocephalus, Lipomyelomeningocele
1
0.2
Spina Bifida Cystica, any Site with Hydrocephalus and Arnold Chiari Malformation
5
1.1
Spina Bifida, any Site with Hydrocephalus of Late Onset
1
0.2
Spina Bifida of any Unspecified Type with Hydrocephalus
1
0.2
Spina Bifida Aperta, without Hydrocephalus
97
21.8
Spina Bifida Cystica, Lumbar, without Hydrocephalus
2
0.5

Spina bifida Cystica, Sacral, without Hydrocephalus
2
0.5
Spina Bifida Other Specified Site without Hydrocephalus
2
0.5
Lipomyelomeningocele
48
10.8
Occipital Encephalocele
9
2.0
Occipital Encephalocele, Anencephaly
1
0.2
Frontal Encephalocele
1
0.2
Parietal Encephalocele
1
0.2
Total
444
100.0
15

Figure 2.3: Distribution of patients born with hydrocephalus

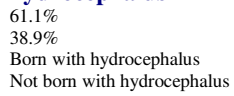
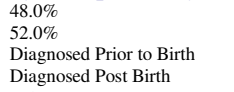


Figure 2.4: Distribution of patients with hydrocephalus by stage of 1st diagnosis



16

Hydrocephalus and antanatal diagnosis

Figure 2.3, shows the percentage of patients who were born with hydrocephalus (61%). Figure 2.4, presents those patients who were diagnosed with hydrocephalus before birth (48%) and those who were diagnosed after birth (52%), with the relevant population being those women who had ultrasound.

Section 3.0: Clinical Data History

Age of repair in days

In figure 3.1 we can see that the majority of patients underwent the primary operation within the first three days of life (53%), (24%) had it between 4-29 days, and 23% had the operation after 30 days of birth.

Figure 3.1: Distribution of patients' age of repair

24.1%
First 3 days of life
4-29 days
>= 30 days

Ventriculoperitoneal shunt insertion

Figure 3.2 presents patients with hydrocephalus who required ventriculoperitoneal shunt insertion, around 61% had VPS inserted, while 39% did not have any.

Figure 3.2: Distribution of patients requiring VPS insertion

60.6%
39.4%
VPS inserted
No VPS

Stage of VPS insertion

Among patients who had VPS inserted 76% had it after repair of the defect, 19% at the repair and only 5% had it before the repair (Figure 3.3). As mentioned before, insertion of VPS after the repair is a standard procedure.

Figure 3.3: Distribution of patients by stage of VPS insertion

75.6%
5.0%
19.4%
At the Repair
After the Repair
Before the Repair

Fractures

Bones usually get stronger through standing and weight bearing. If not used, they can become less thick, fragile and can fracture easily. Patients with **spina bifida** may fracture a leg due to lack of feeling. Table 3.4 shows that the majority of our registry patients did not sustain any fractures 44(88%), 3 patients did not reach developmental milestones of standing yet, to which the “not applicable” code applies.

Table 3.4: Distribution of patients with fractures

Yes
3
6.0
No
44
88.0
Not applicable
3
6.0
Total
50
100.0
17

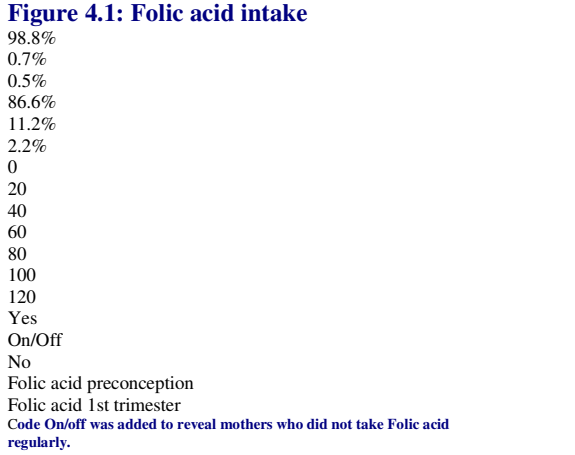
Section 4.0: Risk Factors

Folic acid preconception and third trimester

A large number of mothers in our registry sample did not take folic preconception 407(99%). The first

trimester (348)87% had no folic acid intake as well, with 11% not taking this vitamin regularly (Figure 4.1).

Figure 4.1: Folic acid intake



Code On/off was added to reveal mothers who did not take Folic acid regularly.

Consanguinity

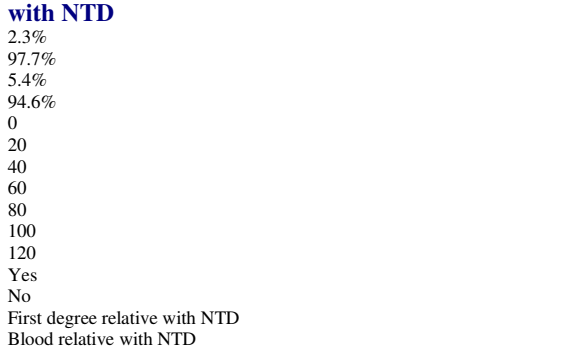
In figure 4.2, 57% of the parents declared they were not related, while 43% declared they were**.**

Figure 4.2: Consanguinity distribution of NTD patients

First degree relative and blood relative with NTD

The vast majority of patients did not have first degree relatives with NTDs (98%), this includes, “Mother, Father, Sibling, Half-sibling and Child”. Moreover a significant percentage of patients (95%) did not have blood relatives with NTDs (figure 4.3).

Figure 4.3: First degree relative and blood relative with NTD



Mothers’ age

In figure 4.4 we notice that a high number of patients were born to mothers between 21-30 years of age 226(52%), followed by 139(32%) for age group between 31-40 years. Furthermore 2 patients were born to mothers over 50 years of age (0.5%).

Figure 4.4: Distribution of age of mothers at birth of child

30
40
50
60
10-14
15-20
21-30
31-40
41-50
> 50
Age groups in years

References

Medical Progress in the Prevention of Neural Tube Defects. Accessed http://www.cdc.gov/ncbddd/bd/mp.htmon 10 February, 2007

Enaw JO, Zhu H, Yang W, Lu W, Shaw GM, Lammer EJ, Finnell RH. CHKA and PCYT1A gene polymorphisms, choline intake and **spina bifida** risk in a California population. BMC Med 2006; 4:36.

Tetsuo Suyama, Kuniyasu Takahashi, Hideo Shibuta, Hiroshi Imaizumi, Shigeru Hirabayashi, Yasuyuki Takakura, Kouichi, Inokuchi. Long results of long rehabilitation in adults with **spina bifida**. J. phys. Ther. Sci. 2000; 12:57-61

APPENDIX A

REGISTERED BY: (Interview)
(Entered)
DATE OF ENTRY:
Neural Tube Defects Registry RAC#: 99 1029E Form NTDR - v3
8 November, 2006
BESC#: 0180/99PD
/ /

KING FAISAL SPECIALIST HOSPITAL AND RESEARCH CENTRE
Registry Core Facility
Biostatistics Epidemiology & Scientific Computing Department
A collaborative effort of Departments of Neurosciences, Pediatrics, Urology, Orthopedic Surgery, Ob. & Gyn.

Neural Tube Defects Registry
Baseline Data Form

P
ATIENT
N
AMEPLATE
INSTITUTION CODE/REGISTRY #:
KFSH&RC MRN:
REGISTRATION DATE:
D D M M Y Y Y Y
DEMOGRAPHIC DATA
Name:
Last
First and Middle
Patient sex:
Male
Female
Unknown
Date of birth:
Nationality:
Saudi
Other Arab

Non Arab
Unknown
D D M M Y
Y Y Y
Telephone #:
Patient's Saudi NID#:
Country Code
Mobile #:
Current Domicile:
If Saudi Arabian
Father's Saudi NID #:
Patrilocality:
Education: Is the patient attending school?
Yes
No
Sometimes
Completed
Disabled
Not Applicable
Education level:
Pre-School
Primary
I
ntermediate
Secondary
College
University
Employment: Is the patient employed?
Yes
No
Not Applicable
If yes:
Full Time
Part time
Irregular Work
Disabled
Mobility:
Non Walker
Household walker
Community walker
Not Applicable
Plurality:
Single
Monozygotic twins
Dizygotic Twins
Unknown
Others, please specify:
Co-twin stillbirth:
Yes
No
Unknown
Family History:
Yes
No if yes, Family Number:
Condition of co-twin:
Normal
Same Defect
Same defects & others
Other defect(s)
Not Applicable
Unknown
Registry # of co-twin:
(Unknown 9999-9999-999, Normal: 8888-8888-888, Blank for N/A)

NEURAL TUBE DEFECT DIAGNOSIS

Condition

BPA Code

ICD-9CM

MACDP

ASSOCIATED DIAGNOSIS (ICD-9-CM CODE)

•

–

•

–

REGISTERED BY: (Interview)
(Entered)
DATE OF ENTRY:
Neural Tube Defects Registry RAC#: 99 1029E Form NTDR - v3
8 November, 2006
BESC#: 0180/99PD
/ /

HISTORY AND DIAGNOSIS
Gestational status at birth:
Pre term
Full term/Post term
Unknown
Sonography Performed:
Yes
No
Unknown
When was NTD diagnosed?
At birth
1st trimester
2nd trimester
3rd trimester
Unknown
Other(s), specify
Was the patient hydrocephalic at birth?
Yes
No
Unknown
Was hydrocephalus diagnosed antenately?
Yes
No
Unknown
Birth Weight:
(gms)
CLINICAL DATA HISTORY
Spina bifida repair:
Yes
No
Unknown
If Yes, date of surgical repair (G) :
D D M M Y
Y Y Y
Hospital of repair:
Wound Complication after the repair:
Yes
No
Unknown
VPS Insertion required?
Yes
No
Unknown
If Yes, at what stage was the VPS inserted?
At the repair
After the repair

Before the repair
Unknown
Not Applicable
VPS complications after insertion:
Yes
No
Unknown
Did the patient sustain any fractures?
Yes
No
Not Applicable
If Yes, specify the site: Upper extremity:
Hand
Arm
Shoulder
Low extremity:
Foot
Leg
Thigh
Pelvis

REGISTERED BY: (Interview)
(Entered)
DATE OF ENTRY:
Neural Tube Defects Registry RAC#: 99 1029E Form NTDR - v3
8 November, 2006
BESC#: 0180/99PD
/ /

Neural Tube Defects Registry

INSTITUTION CODE/REGISTRY #:
KFSH&RC MRN:
PARENTAL DEMOGRAPHICS
Parental consanguinity:
No consanguinity
Related
Unknown
Mother's age at registration:
years
Father's age at registration: years
Patient's mother's history of stillbirths:
Yes
No
Unknown
Patient's mother's residence at conception:
Air conditioned / Air-cooled home
Un-cooled home
Unknown
Not Applicable
History of Intake
Folic Acid
Multivitamins
Yes
Off/on
No Unknown Yes
Off/on
No Unknown
Pre-conception
1st
Trimester
First-degree relative with NTD:
Yes
No
Unknown
Any Blood relative with NTD:
Yes
No
Unknown
First-degree relative
*
with a congenital anomaly:
Yes
No
Unknown
Any Blood relative with a congenital anomaly?
Yes
No
Unknown
(
*
Mother, father, sibling, half-sibling, child)

REGISTERED BY: (Interview)
(Entered)
DATE OF ENTRY:
Neural Tube Defects Registry RAC#: 99 1029E Form NTDR - v3
8 November, 2006
BESC#: 0180/99PD
/ /

KING FAISAL SPECIALIST HOSPITAL AND RESEARCH CENTRE

Registry Core Facility
Biostatistics Epidemiology & Scientific Computing Department
A collaborative effort of Departments of Neurosciences, Pediatrics, Urology, Orthopedic Surgery, Ob. & Gyn.

Neural Tube Defects Registry

Addendum Data Form

P
ATIENT
N
AMEPLATE
INSTITUTION CODE/REGISTRY #:
KFSH&RC MRN:
Follow up DATE:
D D M M Y Y Y Y
Education: Is the patient attending school:
Yes
No
Sometimes
Completed
Disabled
Not Applicable
Education level: :
Preschool
Primary
I
ntermediate
Secondary
College
University
Employment: Is the patient employed:
Yes
No
Not Applicable
If yes :
Full Time
Part time
Irregular Work
Disabled
Mobility:
Non Walker
Household walker
Community walker
Not Applicable
Did the patient sustain any fractures?
Yes
No
Not Applicable
If Yes Specify the site: Upper extremity:
Hand
Arm
Shoulder
Low extremity;
Foot
Leg
Thigh
Pelvis

APPENDIX B

Request for Data from Neural Tube Defects Registry

Name:
ID No.
Position:
Department:
Institution:
MBC No.
Ext.
Date Requested:
Date by which data is required:

1.

Data Requested (specify patient population, time period, etc.)

A. Required Patient Information (specify variables. Use separate sheets if required)

1.
3.
5.
2.
4.
6.

B. Time period

From:
To:

2.

Purpose of the request

Presentation at conference/meeting
Spin-off Research Study
Publication
Patient Care
Other, please specify
Other than Research (specify the reason and provide approval from the Chairman of the Department in your institution)

3.

Is the research study for which the data is requested, approved from Research Advisory Council (RAC)?

Yes
If Yes, provide the RAC Number and attach a copy of the approval memo
No
If No, explain the reasons

4.

If presentation or publication of data is anticipated, identify collaborators and co-authors to be credited:

1.
2.
3.
4.

(Printed Name)
(Signature)

Request Received By:

Confidentiality Statement signed
Registrar, NTD Registry
Dated (DD/MM/YYYY)
Yes
No

For NTD Registry Committee

Request Granted
No
Yes If Yes, date request granted:

Approved by:

Dated (DD/MM/YYYY)
Chairman Registry Committee :

For NTD Registry Use Only

Request Control Number:

Request Completed by:
Date received:
Completion date:

APPENDIX C

King Faisal Specialist Hospital and Research Centre, Riyadh

CONFIDENTIALITY STATEMENT

Name:

ID No.
Position:
Department:
Institution:

I declare that I understand and abide by the rules on confidentiality, security and release of information for users of the
Neural Tube Defect Registry as outlined below.

(Print Name)
(Signature)
(Date)

Rules of Confidentiality, Security and Release of Information for users of Neural Tube Defect Registry Data

1. Data held by Registries Core Facility at Biostatistics, Epidemiology and Scientific Computing Department on patients in Neural Tube Defect Registry
is intended for the purposes of Scientific Research and Statistical Analyses, Healthcare and Hospital Administration support only. The data cannot be used
for any other purpose.
2. Data received from Neural Tube Defect Registry should not be divulged to any person whose name is not specified as a co-user of the data nor
should it be used for any other purposes than that declared in Registry Data Request Form.
3. Proper safeguards should be applied in keeping and destroying the data upon completion of the work/project in order to prevent any breach of confidentiality.
The Chairman of the Registry Committee should be notified immediately of any misuse or loss of data.
4. No patient is to be contacted by a research worker as a result of information supplied by the registry without prior review and consent of the
Registry Committee
5. Any statistics or results of research based on data received from the registry should not be made available in a form which directly identifies individual data
subjects and/or is not covered by the purpose of request specified in the Data Request Form.

Chapter:
Three

Section:
Four

Policy No.
03-04-01

Policy:
Registry Data – Release of Data and/or Information and Result Reporting Policy

Issued:
January 2002

Revised:
October 2006

General:
Data collected by disease registries is directly related to the health care of the patients. This data or any other information
related to the patients' health care is the property of the patient and cannot be released to an unauthorised individual without
prior consent from the registry committee.

Statement:
1. Responsibility of Reporting the descriptive statistics based on the yearly collection of data in the form of an Annual
Report rests upon the registrar of the relevant registry.
2. Any request for release of information / data for research or other purposes should be processed by the Registrar
who is responsible for presenting the request to the Registry Committee or its designee, getting the approval and
downloading the relevant data in ASCII text format. It should be made certain that the whole procedure is in
conformity to the RCF Confidentiality Policy. Registrar is also responsible for maintaining the log of all such

releases of information.

Policy Objective:

To safeguard against unauthorised release of registry information.
To provide a smooth mechanism for the provision of registry data/information to authorised individuals.

Application / Scope:

All registries under RCF.

Monitoring:

Annual

References

RCF IPP # 05-05-01 Confidentiality Policy

Chapter:

Three

Section:

Five

Policy No.

03-05-01

Policy:

Confidentiality Policy

Issued:

January 2002

Revised:

October 2006

General:

Since a disease registry requires the review of significant amounts of data, there is normally a very thorough review of each patient's medical record. All information obtained on patients shall be considered extremely confidential. The actual medical record is the property of the hospital and is kept to document the course of a patient's care and provide communication between all health care professionals for both current and future care of the patient. The actual information contained within the medical record is the patient's property and cannot be released to anyone without proper authorization from the patient, a subpoena, or court order. It is important to stress the strictest confidentiality, as new employees are hired as well as periodic reminders for other employees. RCF members have an obligation to safeguard the confidentiality of personal information maintained in the disease registries. This is governed by ethical and professional codes of conduct. Because of the rapid development of electronic processing of information making sensitive data widely available it is required by the users of sensitive data to ensure they also use common sense when handling data. Different professional and ethical considerations apply depending on the purpose for which the information is used.

Policy Definition:

5.1

Confidentiality

Whilst RCF accepts that great benefits can be made from the information it has collected through disease registries and that medical professionals and hospital management should have ready access to the information they need, it is also important that personal information is kept confidential and that privacy is

respected. Disciplinary action may result from a breach of confidentiality, where a breach of contract can
be proved.

5.2

Principles of Confidentiality

a. The purpose for which data collected by the registry are to be used should be clearly defined.
b. The legal basis of patient registration should be clarified and it should be ensured that all facilities have
legal authority to report the case compulsory or voluntary.
c. All disease registries in the RCF must maintain the same standards of confidentiality as customarily apply to the doctor-patient relationship; this obligation extends indefinitely, even after the death of the patient.
d. Identifiable data may be provided to a clinician for use in the treatment of a particular disease / patient
observing that only the data necessary for the stated purpose are released. Access to patient identifiable information should be on a strict need to know basis. Only those individuals who need access to patient identifiable information should have access to it, and they should only have access to the information items that they need to see. Use the minimum necessary patient identifiable information. Where use of patient identifiable information is considered to be essential, each individual item of information should be justified with the aim of reducing identifiability.
e. The scope of confidentiality extends not only to identifiable data about data subjects and data suppliers, but also to others directly or indirectly identifiable data stored in or provided to the registry.
f. Data on deceased persons should subject to the same procedures for confidentiality as data on living persons.
g. Don't use patient identifiable information unless it is absolutely necessary. Patient identifiable items should only be used if there is no alternative.

h. Everyone should be aware of their responsibilities. Action should be taken to ensure that those handling patient identifiable information, both clinical and non-clinical staff, are aware of their responsibilities and obligations to respect patient confidentiality.
i.
Guidelines for confidentiality apply to all data regardless of storage or transmission media.

Policy Statement:

1. Registrar of each registry is responsible for assuring the confidentiality and security of registry data.
2. The RCF staff should sign, as part of their contract of employment, a declaration that they will not release
confidential information to unauthorised persons. The declaration should remain in force after cessation of
employment. They are also given a copy of the statement. It is essential that the requirements and responsibilities for people working with all the registries, record and databases maintained by Registries
Core Facility (RCF) are clearly defined and understood. This policy outlines the steps that registry database
users must adopt. 'Users' are authorised personnel to access any database. The policy also includes those
staff members who are charged with the responsibility of creation, maintenance and development of registry databases and relevant software in Biostatistics, Epidemiology and Scientific Computing Department.
3. Suitable control of access to the registry, both physical and electronic, and a list of persons, authorised to
enter the registry should be maintained by the Registrar.
4. The Registrar should maintain a list of staff members indicating the nature and extent of their access to
registry data.
5. Notices reminding staff of the need to maintain confidentiality should be promptly displayed.
6. Registries at RCF should provide proof of identity to staff engaged in active patient registration.

7. Identifiable data should not be transmitted by any means (post, telephone or electronic) without explicit
authority from the Head, RCF or staff member to whom such authority has been delegated. Transmission
by telephone should in general be avoided.
8. Registries should consider the use of courier services for confidential data, as well as separating names
from other data for transmission.
9. Precautions should be taken for both physical and electronic security of confidential data sent on magnetic,
optical or electronic media. This could be done by separating identifying information or via encryption of
the identification.
10. Use of computer for confidential data should be controlled for electronic and if possible physical measures
to enhance the security of the data, including use of separate room, passwords, different levels of access to
data, automatic logging of all attempts to enter the system, and automatic closure of sessions after a period
of inactivity.
11. Demonstration of the computer system / database management software should be performed with separate
and fictitious or anonymised data sets.
12. Special precautions should be taken for the physical security of electronic backup media.
13. Expert advice on security against unauthorised remote electronic access should be sought if necessary.
14. Measures should be taken to ensure the physical security of confidential records held on paper or any other
media and to protect such data from corruption.

15. A policy should be developed for the safe disposal of confidential waste.
16. Security procedures should be reviewed at suitable intervals, and consideration should be given to obtaining specialist advice.
17. Any unauthorized release of patient information will be punishable as stated in Oath of Confidentiality.

5.3

Release of Data

a) Release of registry data for research and for healthcare planning is central to the utility of a registry. The registry should develop procedures for data release that ensures the maintenance of confidentiality.
b) The registrar is made responsible to present the request for identifiable data to the Registry Committee
and make recommendations to the committee that the particular request meets the requirement of the law and the registry guidelines on confidentiality. Also the scientific soundness and clinical significance o the project should be judged.
c) In the absence of written consent from data subjects a registry should not release identifiable data on data subjects for the purpose other than research and statistics. National legislation with respect to confidential data should be observed.
d) Physicians should be given access to data needed for the management of their patients if identified as
such and if in accordance with national / institutional regulations after getting approval from the relevant Registry Committee.
e) Provision of own data to the data subject must be given upon request unless institutional / national law
excepts such a release. It is recommended that data subjects be advised to make the request via their own physician.

f) Enquiries from the press should be directed to the Chairman of the relevant Registry Committee or to a
staff member nominated for this purpose.
g) Requests for identifiable data to be used for research should include a detailed justification with a commitment to adhere to the registry's guidelines on confidentiality.
h) Registries should provide a document describing their procedures and criteria for the release of data especially identifiable data to researchers who request access to the data.
i)
If allowed by the institutional and/or national regulations, cross-border transfer of identifiable individual data should only be carried out if required for the conduct of a research project and if the level of protection is satisfactory.

Policy Objective:

The need for a code of conduct in the maintenance of confidentiality in disease registries and the definition of what
should be considered confidential.
The principles of confidentiality including measures to maintain and survey security procedures.
Guidelines for the preservation of confidentiality and for the use and release of registry data in accordance with
these principles.

Application / Scope:

All registries under the umbrella of Registries Core Facility at BESC Department.

Definitions:

Data Subject:
An individual or identifiable natural person, on whom information is processed.
Confidential Data:
For the purpose of this document, any data collected and stored by a disease registry which could permit the identification of an individual patient (data subject) or, in relation to a particular data subject, of an individual physician or institution (data supplier) are considered to be confidential. An identifiable person is one who can be identified directly or indirectly by reference to a reference number or other identifying information such as names, date of birth, national identity number, etc., or to factors specific to his or her physical or physiological, mental, economic cultural or social identity. The collection of unambiguous identifying information on the data subject is necessary to secure quality and use of the registry. The data which, in association with a particular diagnosis, are considered confidential alone, and in combination with other data items are listed below:
1. Names
2. Unique reference numbers
3. Address
4. Full date of birth combined with sex and small area code for place or residence
or death
Security:
Security denotes the measures taken to prevent unauthorised access to the registry data, whether stored on paper or any other media or transmitted by any of these means.
Data Protection:
Includes both the prevention of physical access to the data (security) and the protection of the data to avoid corruption during many years of storage. The term should in this context should not be confused with confidentiality (privacy) the aim of which is to protect the individual from unauthorised disclosures.
Processing of personal data:
Denotes any operation or set of operations that is performed upon personal data whether or not by automatic means such as collection, recording, organization, storage, alteration, retrieval, consultation, use, disclosure by transmission, dissemination or otherwise making available alignment or combination blocking or erasure.
Filing System:

Denotes any means to achieve a structured set of personal data that are accessible according to specific criteria, whether centralised, decentralised or dispersed on a functional or geographical basis.

Informed consent:

Means any freely given specific and informed indication of the wishes of the data subject signifies his or her agreement to personal data relating to him or her being processed.

Policy Review

RCF will review the Confidentiality Policy annually.

References

"Guidelines of confidentiality in population-based cancer registration in the European Union" Adapted by The
European Network of Cancer Registries
North American Association of Central Cancer Registries Policy Statement 99-01: Confidentiality
"Statement of Confidentiality" Arkansas Central Cancer Registry