

DESCRIPTIVE CASES STUDY OF CARE, SURVEILLANCE AND PREVENTION OF BIRTH DEFECTS IN RWANDAN CHILDREN

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ABSTRACT

"According to the World Health Statistics 2008, about 260 000 neonatal deaths worldwide are caused by Congenital anomalies. This figure represents about 7% of all neonatal deaths". In our study, birth accounted Defects for 14.9% out of 581 recruited infants with birth defects (87 cases). In this series, 52.9% were Female whereas 47.1% were male. 13.8% were premature babies (≤ 37 weeks), 74.7% aged < 5 months and 11.5% were infants aged between 5 and 12 months. Polymalformative conditions were the most common Cause of death identified in 21 cases (24%), gastrointestinal birth defects caused death in 15 cases (17%), nervous system in 14 (16%), Cardio-vascular birth defects in 10 cases (12%), Chromosomal abnormalities in 10 cases (12%), musculoskeletal defects in 10 cases (12%), congenital mass in 2 cases (2%), oral defects in 2 cases (2%), congenital skin defect in 1 case (1%), whereas congenital respiratory defect and genitourinary malformations in 1 case each (1%). Over 50% patients died in referral hospitals and 77% died after 24 hours of life.

KEYWORDS: Birth defects - Rwandan children - care - surveillance and prevention.

RESUME

Selon les données statistiques de 2008 de l'Organisation Mondiale de la Santé, environ 260.000 décès néonataux sont causés par des anomalies congénitales. Ceci représente environ 7% de tous les décès en période néonatale.

Dans notre étude, les malformations congénitales représentent 14,9% parmi les 581 enfants nés avec malformation congénitales. Dans cette série, 52,9% étaient du sexe féminin tandis que 47,1% étaient masculins. Les prématurés (≤ 37 semaines de grossesse) représentaient 13,8%; les moins de 5 ans représentaient 74,7% tandis que les enfants âgés entre 5 et 12 mois représentaient une proportion de 11,5% les polymalformations étaient la première cause de décès dans environ 21 cas (24%), tandis que les autres causes de décès étaient malformations gastrointestinales (17%), malformations cérébrales (16%), malformation cardio-vasculaires (12%), anomalies musculo-squelettiques (12%), masses tumorales (2%), malformations de la sphère ORL (2%), les lésions de la peau (1%), et les anomalies respiratoires et de l'appareil génito-urinaire représentaient 1%. Plus de 50% de patients sont décédés à l'hôpital et parmi ces décès 77% sont survenus par les premières 24 heures de la vie.

MOTS CLES: Anomalies congénitales - Rwandan children - Patients Rwandais - Soins - Surveillance et prévention.

INTRODUCTION

"For many centuries birth defects were seen as warnings or divine omens and children with birth defects were often confused with mythological beings. In the eighteenth century there was much interest in maternal impressions. In the mid-nineteenth century it was believed in the English-speaking world that heredity could be altered by external circumstances at any point between conception and weaning. Physicians warned against sexual intercourse under the influence of drugs or alcohol, as they believed that these could affect the constitution of a child. In 1870 the medical profession was criticized for its prevalent belief in the ability of maternal impressions to cause any degree of malformation.

In the late nineteenth century this kind of belief was falling into disrepute because it was linked with magic and "unscientific" thinking. This connection may be a reason the subject was rejected and ignored for so long. As medicine became more scientific, magic was increasingly despised.

By the 1930s there was more serious interest in teratology but, typically, as late as 1937 a popular textbook discussed the diagnosis of fetal abnormalities but not their causes. In 1941, when it was shown that rubella in pregnancy caused birth defects, the peculiarity of the infection was emphasized but it was not taken as a general warning about the vulnerability of fetuses."

By the implementation of medical genetics in different

countries, understanding the birth defects as structural, functional and metabolic disorders present at birth or later and many risk factors has been identified (maternal and environmental factors, drugs in pregnancy and infections). But now birth defects rising up as an important cause of infant mortality even in the developing countries where infant mortality rate has been reduced too much extent.

This retrospective research project, it was then conducted over 12 months in four Rwandan referral hospitals (King Faisal Hospital, Kigali University Teaching Hospital, Butare University Teaching Hospital and Rwanda

Military Hospital). Two districts hospitals in Kigali; Muhima District Hospital and Kibagabaga District Hospital were also included in the study. Patients' files were reviewed and database for birth defects was created. The hospital records were reviewed for demographics; diagnosis, management and outcomes. It focused on understanding the epidemiology of birth defects and mortality in children with birth defects.

LITERATURE REVIEW

Birth defects can be defined as structural or functional abnormalities including metabolic disorders present from birth. The term congenital disorder is considered to have the same meaning and two terms used interchangeably. "It was only in the 20th century that the causes of birth defects were delineated, allowing for their categorization into three broad groups: birth defects originating in the pre-conception period (due primarily to genetic and partly genetic causes); birth defects arising after conception but

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before birth; and birth defects of unknown cause.” According to the March of Dimes (MOD), Global report on birth defects², Worldwide 7.9 million birth occur annually with serious birth defects and 94% of these occur in low and middle

income countries. According to the joint World Health Organization (WHO) and MOD meeting report, birth defects account for 7% of all neonatal mortality and 3.3 million under five deaths¹. In Indian birth defects prevalence varies from 61 to 69.9/1000 live birth. It has been estimated that 70% of birth defects are preventable. Major birth defects including congenital heart defects, neural tube defects (NTDs) and Down syndrome, Hemoglobinopathies, and Glucose-6-Phosphate, dehydrogenase deficiency, cause 20% of infant mortality and are responsible for a substantial number of childhood hospitalizations.

METHODS

Study settings

The study was conducted in four Rwandan referral hospitals (King Faysal Hospital, Kigali University Teaching Hospital, Butare University Teaching Hospital and Rwanda Military Hospital). Two districts hospitals in Kigali, Muhima District Hospital and Kibagabaga District Hospital that are major site for women in labor, were also included in the study.

Study design

This was a retrospective research project, which was conducted over 12 months. The hospital records were reviewed for demographics; diagnosis, management and outcomes.

Study population

Was recruited all children born or admitted in these six hospitals namely four referral hospitals (KFH, CHUK, CHUB, RMH) and two districts hospitals (MDH, KDH); in 2012 who have diagnosed birth defects.

RESULTS

Table 1: Distribution of Birth defects according to the sex

Sex	Number of defects	Percentages
F	320	55%
M	261	45%
Total	581	100

This diagram showed that there is sex-difference in the prevalence of birth defects, female fetuses diagnosed with birth defects represent 55% of all cases of birth defects, and the remaining 45% are male with birth defects. To see how our results are significant we used chi-square test with 95% confidence interval. We found that at 95% CI [0.001-0.002] and P value of 0.017 which is less than 0.05; we concluded that birth defects are more prevalent in female than male.

Table 1bis: Distribution of Birth defects according to the sex

Measures	Birth defects	
Male	261	
Female	320	
P-value	0.017	
95% CI	Lower bound	0.001
	Upper bound	0.002

The subsequent hormonal and physiological distinctions of male and female embryos can explain some sexual differences in frequency of congenital defects said F.I., Boroditsky, R.S., Winter, J.S., Faiman C. (1974).

Table 2: Birth defects distributions in the hospital

Hospital	Total	Percentage
CHUB	86	15%
CHUK	357	61%
KDH	23	4%
KFH	70	12%
MDH	32	6%
RMH	13	2%
Grand Total	581	100%

The Distributions of births defects in Referral and Districts hospital is different, more than 73% of all birth defects were found in referral

hospitals. Kigali University Teaching Hospital is the hospital with many cases, 61% of total birth defects, Butare university Teaching Hospital as the second with 15% of total birth

defects, King Faysal Hospital on the third place with 12% of total birth defects and Rwanda Military Hospitals on the last place with 4% of total birth defects.

Table 3: Birth defects related mortality according to the Hospitals

Hospitals	Dead
CHUB	14
CHUK	41
KDH	3
KFH	18
MDH	11
Grand Total	87

Mortality related to the birth defects is much higher in referral hospitals than in Districts hospitals, in Kigali University Teaching Hospital the mortality associated with birth defects is high compared with others 47%, King

Faysal Hospital as next with 21%. To show how significant our results are, we used Chi-square test, at the 95% CI [0.001-0.002] and P value of 0.001 which is less than 0.05, meaning that mortality is much higher in referral hospitals than districts.

Table 3bis: Birth defects related mortality according to the Hospitals

Measures	Birth defects related mortality	
CHUK	41	
CHUB	14	
KFH	18	
KDH	3	
MDH	11	
P-Value	0.001	
95% CI	Lower bound	0.001
	Upper bound	0.002

Table 4: Birth defects related mortality according to the age

Age	Dead	Percentages	Total
<37wk	12	13.8	12
<5mo	65	74.7	65
5-12mo	10	11.5	10
Grand Total	87	100	87

This diagram is describing the mortality related to the birth defects according to the ages, the term infants with ages less than five months had high mortality 75% compared with premature babies 14% and those with ages of 5-12months 11%. By using Chi-Square test at 95% we found a CI [0.0001-0.0002] and P value of 0.0001 which is less than 0.05 we ended by concluding that our results are significant high mortality is in term babies with age between 5-12months.

Measures	Birth defects	
<37wks	12	
<5mo	65	
>5mo	10	
P-Value	0.0001	
95% CI	Lower bound	0.0001
	Upper bound	0.0002

Table 5: Birth defects related mortality according to the sex

Sex	Dead	Percentages	Total
M	46	53	46
F	41	47	41
Grand Total	87	100	87

53% of all children died from birth defects are male and the remaining 47% are female. To see how significant these results are. We did Chi-square test and at 95% [0.57-0.128] and P value of 0.105 which is greater than 0.05, we concluded that the association between sex and outcome or death is not statistically significant.

Measures	Birth defects	
Male	46	
Female	41	
P-value	0.105	
95% C	Lower bound	0.57
	Upper bound	0.128

Table 6: Birth defects classes and related mortality

	Dead	Percentage	Grand Total
Chromosomal Abnor	10	12%	10
Congen Cardiovas Def	10	12%	10
Congen Gastrointest Def	15	17%	15
Congen Genitourinary Def	1	1%	1
Congen Musclesk Def	10	12%	10
Congen Nervous Def	14	16%	14
Congen oral defect	2	2%	2
Congen Polymalformation	21	24%	21
Congen Resp Def	1	1%	1
Congen Skin Def	1	1%	1
Congenital Mass	2	2%	2
Grand Total	87	100%	87

Polymalformative conditions were the most common cause of death identified in 21 cases (24%), gastrointestinal birth defects caused death in 15 cases (17%), nervous system in 14 (16%), Cardio-vascular birth defects in 10 cases (12%), Chromosomal abnormalities in 10 cases (12%), musculoskeletal defects in 10 cases (12%), congenital mass in 2 cases (2%), oral defects in 2 cases (2%), congenital skin defect in 1 case (1%), whereas congenital respiratory defect and genitourinary malformations in 1 case (1%), respectively.

DISCUSSION

All children born in 2012, who consulted one of Rwandan referral hospitals (Kigali University Teaching Hospitals, Butare University Teaching Hospitals, King Faysal Hospital and Rwanda Military Hospital) or one of these two

districts hospitals in Kigali (Muhima District Hospital and Kibagabaga District Hospital), their files were reviewed and 581 born in 2012 with Birth defects, Among these 87(14.9%) died when they were less than five months and more than 50% died in referral hospitals.

The children with birth defects who were born out of 2012 were excluded from the study. "More than 90 percent of all infant with serious birth defects are born in middle and low income countries. Because most of these countries don't have adequate services to care for infant and children with birth defects, many of them will die young." (March of Dimer global report on birth defects, 2004).

In this retrospective case study more than 80 percent of all birth defects were born in referral hospitals and among them, more than 50 percent died in referral hospitals.

Even though Rwandan referrals hospitals can diagnose some structure birth defects and manage them surgically, many of these defects are diagnosed too late because of neglected prenatal care of the pregnant women or incompetent health care provider for diagnosis of congenital malformation in the early stage, thus this increases the mortality in children with birth defects especially in referral hospitals. Children with functional problems like thalassemia, sickle cell diseases, congenital hypothyroidism, and glucose-6-deficiency phosphate dehydrogenase are more at high risks of dying before diagnosis compared to the children with structure defects and chromosomal abnormalities.

We are still considering poverty, maternal age at conception, proportion of unplanned pregnancies and no antenatal care, maternal nutritional status and exposure to teratogens before and after conception as the basic risks factors that are increasing the number of births defects in Rwandan pediatric children.

In this study the mortality related to birth defects was high in infants under five months 75% and the ratio for male to female is 1.12.

Several studies done on birth defects showed that the mortality is high in children under five years, As it was reported in Global report on birth defects done by March of Dimes in 2004 "At least 3.3 million children less than 5 years of age die annually because of serious birth defects and the majority of those who survive may be mentally and physically disabled for life", but this study proved that is high in children under five months, the factors that are in favor of high mortality under five months children of life compared to those studies is that our study was limited to the children of one year and in our hospitals diagnosing birth defects and management for those children is inadequate to make them live longer.

In this study, 581 children was born in 2012 with birth defects, and congenital nervous defects are the most common in 138(24%) and the second is Congenital gastro-intestinal defects in 124(21%) then Polymalformative syndromes 86(15%), musculoskeletal defects 64(11%), cardiovascular defects in 49(8%) infants. But according to the literature cardiovascular defects, neural defects, Chromosomal defects and Gastro-intestinal defects are the most common.

The findings from our studies are supporting other

studies done means that as the world facing the problem of burden of mortality related to the birth defects our country too should know that and prepare the strong preventive measures.

Many facts can support our results, like the physical manifestation of birth defects; as the defects manifest physically the more the mothers become worried and consult the hospitals in the early ages of the infants. In our study congenital nervous defects are the most common where hydrocephalus (33%), myelomeningocele (28%) and spinal bifida (24%) in infants less than one year old because these are easily detected even in prenatal care on ultrasound. The second are the gastro-intestinal defects where hypertrophic pyloric stenosis (20,1%), imperforated anus (20%) and omphalocele (19%) are the most common.

The early age manifestation and easily diagnosis of some birth defects can make a small deference in the order of prevalence of birth defects between developing and developed countries. In developing countries, as the defects manifest physically and easily diagnose like spinal bifida on trans-abdominal or trans-vaginal ultrasound the more prevalence and the cheaper the test is, on the other hand, in developed countries where screening of many birth defects is mandatory, the prevalence will depends on teratogens the mother exposed to during the early pregnancy, the number of children born during that period and the quality of diagnostic tools, but if both developed and developing countries are in the same settings the birth defects prevalence order will be the same.

"Major birth defects are diagnosed for 3%–4% of infants in their first year of life. Of the 100,000–150,000 infants born with a major birth defect each year (3) approximately 6,000 die during their first 28 days of life, and another 2,000 die before reaching their first birthday. The remaining 92,000–142,000 children who survived beyond the age of 1 year are affected by birth defects to various degrees."

"EUROCAT (European Surveillance of Congenital Anomalies) is the network of population-based registers of congenital anomaly in Europe, with a common protocol and data quality review, covering 1.5 million annual births in 22 countries. EUROCAT recorded a total prevalence of major congenital anomalies of 23.9 per 1,000 births for 2003-2007. 80% were live births. 2.5% of live births with congenital anomaly died in the first week of life. 2.0% were stillbirths or fetal deaths from 20 weeks gestation. 17.6% of all cases were terminations of pregnancy following prenatal diagnosis (TOPFA)".

In our study for a total of 581 infants born in 2012 were diagnosed with birth defects and 97% were live birth and 3% died less than 32 weeks of pregnancy. Among 97% who are live birth, 11% died at less than 5 months and 2% died between 5 months and 12 months.

Like other studies done about the prevalence of birth defects, distribution according to the age and sex, diagnosis and management, and mortality related to birth defects were all proved that; the birth defects are becoming a one of major public health problem and is important cause of infants mortality especially in low and middle income countries where 94% of all children die from birth defects were found (March of Dimes global report on birth defects, 2004). As the results from our study done in Rwandan referral hospitals and two Kigali district hospitals, they are

supporting other findings about birth defects; we have to stand for applying preventives measures in order to protect our newborn from birth defects.

CONCLUSION

The birth defects have never been considered especially in developing countries that is why nowadays they are

becoming one of the major causes of neonatal deaths around the world, but is more significant in those countries. We should stand for prevention of birth defects in middle and low incomes countries as we do for HIV, TB, Malaria and other diseases.

REFERENCES

1. Arnold Christianson, Christopher P. Howson, Bernadette Modell.(2006). March of Dimes Birth Defects Foundation. White Plains, New York.
2. Diseases control priorities project. (Dec 2008).Controlling Birth Defects: Reducing the hidden Toll of Dying and Disabled Children in Low-Income Countries.
3. Lowell E. Sever, Ph.D. June 2004Guidelines for ConductingBirth Defects Surveillance
4. Lopez, A.D., et al., Global and regional burden of disease and risk factors, 2001: systematic analysis of population health data. Lancet, 2006.367(9524)
5. Report WHO consultation on community genetics in low and middle countries. Community genetic services.Geneve, Switzerland (2010). WORLD HEALTH ORGANIZATION CONSULTATION (WHO).2010.
6. Resolution WHA63.17. Birth Defects. In: Sixty-third World Health Assembly,Geneva, 21 May 2010.Geneva, World Health Organization, 2010.
7. International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) WHO Collaborating Center. Birth Defect Prevention: Global Issues.WHO, Geneva, 16 January 2012: Hosts, Dr. Mario Meriardi, Dr. JP Pena-Rosas
8. World health statistics 2008.Birth defects:Report by the Secretariat. World Health Organization, Geneva, 2008.Executive Board 125/7(EB125/7)-may 2009.
9. Resolution A63/10.Birth Defects: Report by Secretariat. In: Sixty-third World Health Assembly, Geneva,1 April 2010. Geneva, World Health Organization,2010.
10. Community Genet 2002; PREVENTING CONGENITAL ANOMALIES IN DEVELOPING COUNTRIES. New York, NY 10003, USA. Victor B. PenchaszadehDivision of Medical Genetics
11. Inequalities in Infant Mortality Project Briefing Paper 4, the contribution of Congenital anomalies to infant mortality.Jennifer J Kurinczuk, Jennifer Hallowell,Patricia A Boyd, Laura Oakley, Peter Brocklehurst, Ron Gray, National Perinatal Epidemiology Unit, University of Oxford.June 2010
12. World health organization. Management of birth defects and haemoglobin disorders: Report of a Joint WHO-March of Dimes meeting. Geneva, Switzerland, Geneva: WHO; 2006.
13. Christianson AL, Howson CP, Modell B. White Plains. New York, USA: March of Dimes Birth Defects Foundation; 2006. [Last accessed on 2012 Feb 24]. March of dimes global report on birth defects: The hidden toll of dying and disabled children. Available from: <http://www.marchofdimes.com> .
14. Diav-Citrin O, Koren G. Nausea and Vomiting of Pregnancy: State of the Art 2000. Toronto, Ontario, Canada: The Motherisk Program, the Hospital for Sick Children; 2000. Human teratogen: A critical evaluation.
15. International Institute for Population Sciences (IIPS); Macro International National Family Health Survey (NFHS-3), 2005-06. Vol. 1. India, Mumbai: IIPS; 2007.
16. Government of India. Sample registration system of India. Office of Registrar General of India. Ministry of Home Affairs. GOI. 2007 Oct;42:1-6.
17. Ghosh S. Down syndrome in India. Br Med J. 1967;3(29):309-1.
18. World Health Organization. Country profile on reproductive health in Bangladesh. Geneva: WHO; 2003.