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Prevalence of Newborn Blood Disorders at Maternity and Children's Hospital, Ksa (1437 H)

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Abstract: *This student project was conducted to assess the prevalence of blood disorders among newborn at Maternity and Children's Hospital, Kingdom of Saudi Arabia. It sought to answer the following questions: (1) what is the profile of the newborn? 2) How many newborn were positive for the blood disorder? 3) What is the percentage of the newborn blood disorder were managed at maternity and children's hospital? The participants of the study were the 6,047 newborn and documentary research design was used where the data were gathered from the Pediatric Clinic of Maternity and Children's Hospital. Frequency and percentage were used to treat the data. The results revealed that majority of the newborn in Maternity and Children's Hospital in Hail City were Saudi, female and first birth order. The newborn blood disorders detected were Congenital Adrenal Hyperplasia, Maple Syrup Urine Disease, Congenital Hypothyroidism and Galactosemia and the most prevalent were Congenital Hypothyroidism and Galactosemia. The managed newborn blood disorder was Congenital Hypothyroidism.*

Keywords: *Newborn blood disorder, Managed blood disorder, Congenital Adrenal Hyperplasia, Maple Syrup Urine Disease, Congenital Hypothyroidism, Galactosemia.*

I. Introduction

Newborn screening is the practice of testing every newborn for certain harmful or potentially fatal disorder that isn't otherwise apparent at birth [(1)]. And for those babies with the conditions, it allows doctors to start treatment before some of the harmful effects happen. and in most cases, while you are still in the hospital all it takes is a few drops of blood and a simple hearing test, [(2)] Other disorders that screening can detect include problems with hormones or the blood [(1)]. In general, metabolic and other inherited disorders can hinder an infant's normal physical and mental development in a variety of ways [(3)]. Newborn screening is ideally done immediately after 24 hours from birth. Babies with positive results must be referred at once to a specialist for confirmatory testing and further management. Should there be no specialist in the area; the NBS secretariat office will assist its attending physical. [(4)] The disorders tested for newborn screening are: Congenital Hypothyroidism (CH) Congenital Adrenal Hyperplasia (CAH), Galactosemia (GAL), Phenylketonuria (PKU) Glucose-6-Phosphate-Dehydrogenase Deficiency (G6PD Def.),Maple Syrup Urine Disease (MSUD)[(4)] Congenital Hypothyroidism (CH) is a condition of thyroid hormone deficiency present at birth. Approximately 1 in 4000 newborn infants has a severe deficiency of thyroid function, while even more have mild or partial degrees. If untreated for several months after birth, severe congenital hypothyroidism can lead to growth failure and permanent intellectual disability. Treatment consists of a daily dose of thyroid hormone (thyroxine) by mouth. Because the treatment is simple, effective, and inexpensive, nearly all of the developed world practices newborn screening to detect and treat congenital hypothyroidism in the first weeks of life. [(5)]

Congenital Adrenal Hyperplasia (CAH) is an endocrine disorder that causes severe salt loss, dehydration and abnormally high levels of male sex hormones in both boys and girls. If not detected and treated early, babies with CAH may die within 7-14 days.[(4)] Galactosemia (GAL) is a condition in which babies are unable to process galactose, the sugar present in milk. Accumulation of excessive galactose in the body can cause many problems, including liver damage, brain damage and cataracts. [(4)] Phenylketonuria (PKU) is a rare condition. A phenylketonuria (PKU) test is done to check whether a newborn baby has the enzyme needed to use phenylalanine in his or her body. Phenylalanine is an amino acid that is needed for normal growth and development. If a baby's body does not have the enzyme that changes phenylalanine into another amino acid called tyrosine, the phenylalanine level builds up in the baby's blood and can cause brain damage, seizures, and intellectual disability.[(6)] Glucose-6Phosphate Dehydrogenase Deficiency (G6PD def) deficiency is a condition where the body lacks the enzyme called G6PD.Â Babies with this deficiency may have hemolytic anemia resulting from exposure to oxidative substances found in drugs, foods and chemicals.[(4)]It is an inherited condition of the blood. These exposures can trigger the red blood cells, which carry oxygen around the body, to break down prematurely [(7)].

Maple Syrup Urine Disease (MSUD) MSUD is a genetic metabolic disorder resulting from the defective activity of the enzyme branched chain alpha-keto-acid dehydrogenase complex. Accumulation of the branched chain amino acids are toxic to the brain.[4]

The effect on the newborn if the blood disorders are not screened includes the following: Congenital Hypothyroidism (CH) and Phenylketonuria (PKU) can cause severe mental retardation, Congenital Adrenal Hyperplasia (CAH) and Maple Syrup Urine Disease can lead to death, G6PD deficiency leads to severe anemia and kernicterus, Galactosemia (GAL) can cause death or cataracts. However, the newborn will be alive and normal if the blood disorders are screened and managed. .[4]

Ideally, all newborn should be screened for all disorders for which effective treatment is available. These disorders can be grouped into 5 categories: amino acid metabolism disorders, organic acid metabolism disorders, fatty acid oxidation disorders, hemoglobinopathies, and others (namely congenital. Hypothyroidism, cystic fibrosis, hearing loss, congenital adrenal hyperplasia. Galactosemia and biotinidase deficiency).[8]

DNA-based Techniques : DNA based techniques are introduced recently to newborn screening programs, In Saudi Arabia it is available in King Faisal Specialist Hospital and Research center and other few places as well. The tests are designed to test many different conditions in one blood sample, and have relatively rapid results in spite of it is accuracy, however it needs trained experts to operate it. DNA -based test added great value for testing. Together with the advanced technology like MS/MS, IEF, and HPLC for hemoglobinopathies variants, DNA arrays used in sequencing, and labeled bead technologies are also used with great success. [9]

1.1 Objectives and Statement of the Problem:

The objectives of this study were to determine the blood disorders of newborn and to determine what blood disorders were managed through treatment.

Statement of the problem: 1) what is the profile of the new born? 2) How many new born were positive for the blood disorders? 3) What is the percentage of the new born blood disorder were managed at Maternity and Children's Hospital?

II. Methodology

2.1 Participants

The participants of the study were 6,047 newborn in Maternity and Children Hospital Hail City, Saudi Arabia covering the year 1437 Higgry. The data were taken from pediatric clinic of the said hospital.

2.2 Materials

The materials and or instruments used for the determination of blood disorders of newborn were the following: cotton, alcohol swab, sterile lancet, gauze, filter paper for newborn screening.

2.3 Procedure

The primary goal of this standard is to ensure the quality of blood spots collected from newborns. Poor quality specimens place an unnecessary burden on the screening facility, because unnecessary trauma to the infant and anxiety to the infant's parents, potentially delay the detection and treatment of the affected infant, and may contribute to a missed or late diagnosed case. If the Newborn Screening Program receives an unacceptable specimen, program staff requests a repeat sample from the birthing center. Blood collected from the heel is the standard for newborn screening. The medial and lateral parts of the underfoot are preferred. Warm site with soft cloth, moistened with warm water up to 41°C, for three to five minutes. Cleanse site with alcohol prep. Wipe dry with sterile gauze pad. Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another large blood drop to form. Lightly touch filter paper to large blood drop. Allow blood to soak through and completely fill circle with single application to large blood drop. Fill all required circles with blood. Apply blood to one side of filter paper only.

The specimen should be allowed to air dry in a horizontal position for at least 3 hours before mailing. Neither side of the blood spots may touch a surface. Specimens are to be kept away from direct sunlight and heat sources during the drying process. Heat must not be used to facilitate drying. All specimens must be sent to the laboratory at the day of collection. Specimens should not be placed in plastic bags prior to mailing. The newborn blood specimens were sent to Riyadh King Saud Hospital for the screening test.

2.4 Statistical Analysis Tools

Frequency and Percentage were used to treat the data that were gathered.

III. Results

Table 1.1 Profile of the Newborn According to Nationality

Nationality	Frequency	Percentage
Saudi	6,047	100%
Non-Saudi	0	0%
TOTAL	6,047	100%

Table 1.1 presents the result on the percentage of profile of the newborn based on nationality. It tells us that 100% of the newborn were Saudi and 0% non-Saudi. This means that majority of the newborn in Maternity and Children's Hospital were Saudi.

Table 1.2 Profile of the newborn (Gender)

Gender:	Frequency	Percentage
Female	3,550	58.71%
Male	2,497	41.29%
Total	6,047	100%

Table 1.2 presents the result on percentage profile of the newborn based on gender. It tells us that 58.71% of the newborn were female and 41.29% were male. This means that majority of the newborn were female.

Table 1.3 Profile of the Newborn According to Birth Order

Birth order:	Frequency	Percentage
1st	2,200	36.38%
2nd	740	12.24%
3rd	1360	22.49%
4th	645	10.66%
5th	659	10.90%
6th	443	7.33%
Total:	6,047	100%

Table 1.3 presents the result on percentage of profile of the newborn according to birth order. It tells us that 36.38% belonged to the 1st, followed by 3rd birth order with 22,49 % , 2nd birth order with 12.24 , 5th birth order with 10.90% , 4th birth order with 10.66 % and 6th birth order with 7.33% . This means that majority of the newborn were in the first birth order and the least were the 6th birth order.

Table 2 Percentage Result on Newborn Blood Disorders

Newborn Blood Disorders	N	Negative		Positive	
		Frequency	Percentage	Frequency	Percentage
Congenital Hypothyroidism	6,047	6,040	99.83 %	7	0.12 %
Congenital Adrenal Hyperplasia	6,047	6,035	99.80 %	12	0.20 %
Galactosemia	6,047	6,040	99.83 %	7	0.12 %
Maple Syrup Urine Disease	6,047	6,035	99.80 %	12	0.20 %

Table 2 presents the percentage result on newborn blood disorder detected at Maternity and Children's Hospital. It tells us that 0.20% were Congenital Adrenal Hyperplasia and Maple Syrup Urine Disease, followed by Congenital Hypothyroidism, and Galactosemia with 0.12 %. This means that majority of the newborn blood disorders detected were Congenital Adrenal Hyperplasia and Maple Syrup Urine Disease and the least were Congenital Hypothyroidism and Galactosemia.

Table 3 Percentage Result of Managed Newborn Disorders

	Congenital Hypothyroidism		Congenital Adrenal Hyperplasia		Galactosemia		Maple Syrup Urine Disease	
	Frequency	Percentage	Frequency	Percentage	Frequency	Percentage	Frequency	Percentage
Managed Alive	4	57%	2	17%	3	43%	4	33%
Not Managed Alive	0	0%	3	25%	0	0%	3	25%
Death	3	43%	7	58%	4	57%	5	42%
Total	7	100 %	12	100 %	7	100 %	12	100 %

Table 3 shows the percentage result of **managed** newborn blood disorders. This tells us that most of the cases on congenital hypothyroidism blood disorder for newborn were managed as indicated by 57 %, followed by Galactosemia (43 %), Maple Syrup Urine Disease (33 %) and Congenital Adrenal Hyperplasia (17 %). Moreover, congenital adrenal hyperplasia and maple syrup urine disease results revealed that 25 % of the cases were not managed but the newborn were alive. Percentage of death is highest in Congenital Adrenal Hyperplasia (58 %), followed by Galactosemia (57%), Congenital Hypothyroidism (43 %) and Maple Syrup Urine Disease (42%).

This means that most of the newborn with Congenital Hypothyroidism blood disorder were managed , therefore , the newborn were alive and normal .However , the highest incidence of death were the newborn with Congenital Adrenal Hyperplasia blood disorder.

IV. Discussion

Newborn screening (NBS) is the practice of testing every newborn for treatable genetic, metabolic, endocrinological and haematological disorders.[1] Newborn screening test will prevent developmental delay, mental retardation and death of infants.[10] Some initial symptoms characteristic of classic MSUD are: lethargy, poor appetite ,weight loss, weak sucking ability, irritability distinctive maple sugar odor in earwax, sweat, and urine, irregular sleep patterns alternating episodes of hypertonia (muscle rigidity) and hypotonia (muscle limpness),high-pitched cry. [11] Some initial symptoms characteristic of classic CAH poor feeding lethargy, vomiting, weight loss, dehydration, rapid heart rate.[12] Symptoms CH puffy face, swelling around the eyes, poor feeding , constipation , yellowish skin (jaundice), Large soft spot on the head that is slow to close.[13]

A review on the current experiences with newborn screening in the Middle East and North Africa region. The population in the region is about 400 million, with high birth rate and an estimated 10 million newborns per year. The majority of the population is of the Islamic faith and mostly Arab. The population is characterized by a high consanguinity (25–70%) and a high percentage of first-cousin marriages. Haemoglobin disorders, inherited metabolic disorders, neurogenetic disorders and birth defects are relatively common among the population. There is a rather slow progress in developing and implementing preventive genetic programmes owing to legal, cultural, political and financial issues. Although research spending is rather soft in the region, there are numerous pilot studies that highlighted the high incidence of genetic defects and the need for newborn screening programmes. Currently, there are only four countries that are executing national newborn screening but they vary from one disease to 23 and coverage is not complete. The region needs to take big steps towards developing national strategies for prevention and should learn from experiences of regional and international screening programmes [13]. Over a 12-year period (1983 to 1994), 11 cases of classical galactosaemia were identified in the Saudi Aramco Medical Services Organization (SAMSO).

The result of the study as reflected in Table 2, on newborn blood disorder coincides with the findings of Saudi Aramco medical services were an incidence on galactosaemia of 12 cases per 100,000 live births in the Easter Province of Saudi Arabia. A higher proportion of infants presented with infections compared to reports in the literature. Screening for galactosaemia is highly recommended in this high-risk population. Moreover, in a study conducted in King Faisal Specialist Hospital and Research Center (the largest referral hospital for congenital and metabolic diseases in the country), Al-Odaib ET al.reported the major Inborn Error of metabolism seen at a pediatrics clinic from (1998-2002). A total of 96,015 newborns were tested from January 1990 - December 2007, found (26) cases of primary congenital hypothyroidism (CH); (6) cases of transient hypothyroidism; (13) cases of congenital hypothyroidism. [9]

V. Conclusion

Majority of the newborn in Maternity and Children's Hospital in Hail City, Saudi Arabia were Saudi , female and belonged to the first birth order. The newborn blood disorders detected were Congenital Adrenal Hyperplasia, Maple Syrup Urine Disease, Congenital Hypothyroidism and Galactosemia. The most prevalent newborn blood disorder detected were Congenital Hypothyroidism and Galactosemia. Although some of the newborn blood disorder cases were managed, however, there were still cases where the blood disorders were not managed yet the newborn were alive. Incidence of death were also noted for all newborn cases with the highest incidence on Congenital Adrenal Hyperplasia. It is recommended for all hospitals in the kingdom to monitor the treatment of babies who were screened and found out to be positive for the blood disorders most especially in cases such as Congenital Adrenal Hyperplasia which resulted to a high mortality rate among newborn.

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References

[1]. http://kidshealth.org/en/parents/newborn-screening-tests.htm
[2]. http://babysfirsttest.org/newborn-screening/screening-101
[3]. http://kidshealth.org/CMNuA/en/parents/newborn-screening-tests.html?WT.ac=clk_frommob
[4]. http://www.newbornscreening.ph/index.php?option=com_content&view=section&layout=blog&id=3&Itemid=60
[5]. https://en.wikipedia.org/wiki/Congenital_hypothyroidism
[6]. http://www.webmd.com/parenting/baby/phenylketonuria-pku-test#1
[7]. http://www.babysfirsttest.org/newborn-screening/conditions/glucose-6-phosphate-dehydrogenase-deficiency
[8]. https://www.google.com.sa/url?sa=t&rct=j&q=&esrc=s&source=web&cd=1&ved=0ahUKEwiK0KHiu7_QAhXE0hoKHZgEBFgQFggbMAA&url=http%3A%2F%2Fsmj.org.sa%2Findex.php%2Fsmj%2Farticle%2FviewFile%2F5916%2F3690&usg=AFQjCNF7HiwOELvx7Nq7keGibw_PV6n2Lg
[9]. https://www.ukessays.com/essays/health-and-social-care/newborn-screening-in-saudi-arabia.php
[10]. http://hamdanjournal.org/journal/index.php?journal=HAMDAN&page=article&op=view&path%5B%5D=192
[11]. http://www.healthline.com/health/maple-syrup-urine-disease#Types2
[12]. https://www.newbornscreening.on.ca/en/disease/galactosemia
[13]. Moammar H, Ratard R, Cheriyan G, Mathew P. Incidence and features of galactosemia in Saudi Arabs. J Inherit Metab Dis. 1996; 19:331–4.