



Newborn Screening in Bisha region, Saudi Arabia: A 14 Years Retrospective Study on the Incidence of Inborn Errors of Metabolism and Comparisons with Regional and Global Data

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Abstract

Background: Newborn screening (NBS) is a population-based public health program designed to identify newborns with serious but treatable inborn error of metabolism disorders before the onset of clinical symptoms. Early diagnosis through NBS enables timely intervention, thereby reducing morbidity, mortality, and long-term disability. Saudi Arabia has one of the highest reported incidences of inborn error of metabolism and endocrine disorders worldwide, largely attributed to the high prevalence of consanguineous marriages.

Objective: To determine the incidence of inborn error of metabolism and endocrine disorders detected through the Saudi National Newborn Screening Program in the Bisha region over a 14-year period and to compare the findings with national and international incidence data.

Methods: A retrospective observational study was conducted using data collected between January 1, 2006, and December 31, 2019. During the study period, 73,571 newborns, representing 97.3% of all live births (73,571/75,600), underwent newborn screening in Ministry of Health hospitals in the Bisha region. Heel-prick dried blood spot samples were collected between 24 and 72 hours after birth and analyzed at the National Health Laboratory using tandem mass spectrometry and immunofluorometric assays. Only laboratory-confirmed cases were included in the analysis. Disease incidences were calculated and compared with previously published data from other Saudi regions and international populations.

Results: Among 73,571 screened newborns, 78 were diagnosed with inherited metabolic or endocrine disorders, corresponding to an overall incidence of approximately 1 in 943 live births. Argininosuccinate lyase deficiency (ASLD) was the most frequently detected disorder (19 cases; incidence 1: 3,872), congenital hypothyroidism (19 cases; 1:3,872), and Propionic acidemia/methylmalonic acidemia (PA/MMA) (9 cases; 1:8,174). Compared with published Saudi and international data, Bisha region has high overall incidence, ASLD and PA/MMA were markedly higher in the Bisha region.

Conclusion: The Bisha region demonstrates high overall incidence of 1:943 and a substantially higher incidence of ASLD and PA/MMA than what has been reported from other regions and internationally. This may reflect the influence of consanguinity and founder mutations within the local population. These findings emphasize the importance of maintaining comprehensive newborn screening services and support the implementation of enhanced genetic counseling, premarital screening, and targeted carrier screening in high-risk communities.

Keywords: Newborn Screening; Inborn Errors of Metabolism; Incidence; Tandem Mass Spectrometry; Consanguinity; Bisha; Saudi Arabia

Abbreviations: ASLD: Argininosuccinate Lyase Deficiency; BTD: Biotinidase Deficiency; CAH: Congenital Adrenal Hyperplasia; CH: Congenital Hypothyroidism; GA-1: Glutaric Acidemia Type 1; GALT: Galactosemia; IVA: Isovaleric Acidemia; VLCAD: Very Long-Chain Acyl-CoA Dehydrogenase Deficiency; 3-MCC: 3-Methylcrotonyl-CoA Carboxylase Deficiency; MMA: Methylmalonic Acidemia; MSUD: Maple Syrup Urine Disease; PA/MMA: Propionic Acidemia/Methylmalonic Acidemia; PKU: Phenylketonuria; IEM: Inborn Error of Metabolism; NBS: Newborn Screening.

Introduction

Newborn screening (NBS) is one of the most successful public health initiatives for the early detection of serious but treatable congenital disorders. By identifying affected infants before the onset of clinical manifestations, NBS enables timely intervention, thereby reducing mortality, preventing irreversible neurological impairment, and improving long-term health outcomes [1-7]. Since the introduction of tandem mass spectrometry (MS/MS), the scope of newborn screening has expanded considerably, allowing simultaneous detection of numerous inborn errors of metabolism (IEM) and endocrine disorders from a single dried blood spot specimen collected during the first few days of life [8,9].

Inborn error of metabolism comprises a heterogeneous group of predominantly autosomal recessive diseases caused by defects in metabolic pathways involving amino acid metabolism, organic acid metabolism, fatty acid oxidation, carbohydrate metabolism, and cofactor synthesis. Although each disorder is individually rare, their collective incidence is clinically significant, particularly in populations with a high prevalence of consanguineous marriage, especially in the southwestern regions such as Asir and Bisha [10-13]. Early diagnosis through newborn screening permits the initiation of dietary modification, pharmacological therapy, hormone replacement, or other disease-specific interventions before irreversible complications develop.

The Saudi National Newborn Screening Program was established in 2005 by the Ministry of Health and has progressively expanded to provide nationwide screening for selected inherited metabolic and endocrine disorders [1-2,14]. During the study period, the screening panel included 17 disorders, comprising amino acid disorders, organic acidemias, fatty acid oxidation disorders, ketogenesis and ketolysis defects, galactosemia, congenital hypothyroidism, congenital adrenal hyperplasia, and biotinidase deficiency. Screening is performed using tandem mass spectrometry for inborn error of metabolism disorders and immunofluorometric assays for endocrine disorders, with confirmatory testing undertaken for all screen-positive newborns.

Saudi Arabia reports one of the highest incidences of IEM worldwide. This has largely been attributed to the high prevalence of consanguineous marriage, founder variants, and large family size within many communities [10,11]. National newborn screening studies have demonstrated considerable regional variation in genetics and disease incidence, suggesting that local epidemiological data are essential for healthcare planning, resource allocation, and the development of targeted preventive strategies. However, published data from the Bisha region remain limited despite the long-standing implementation of the national screening program.

The Bisha region is located in the southwest of Saudi Arabia and is served by five Ministry of Health hospitals, with King Abdullah Hospital functioning as the regional tertiary referral center. Approximately 5,000–6,000 live births occur annually within the region. Because of the demographic characteristics of the local population, regional evaluation of IEM disorders is particularly important for understanding disease burden and guiding preventive healthcare policies.

The present study aimed to determine the incidence of inborn error of metabolic and endocrine disorders detected through the Saudi National Newborn Screening Program in the Bisha region over a 14-year period and to compare the regional incidence with previously published data from other regions of Saudi Arabia and international populations.

Methods

Study Design and Setting

Institutional approval was obtained from the ethical committee of King Abdullah Hospital. This is retrospective observational study analyzed data collected through the Saudi National Newborn Screening (NBS) Program in the Bisha region, Kingdom of Saudi Arabia [1,13], over a 14-year period from 1 January 2006 to 31 December 2019. The Bisha region includes five Ministry of Health (MOH) hospitals, with King Abdullah Hospital serving as the regional tertiary referral center. Approximately 5,000–6,000 live births occur annually within the region.

Study Population

All live-born infants delivered in MOH hospitals in the Bisha region during the study period were eligible for newborn screening. A total of 73,571 newborns, representing 97.3% of the 75,600 live births, underwent screening and were included in the analysis. Newborns who were not screened or whose screening results were incomplete or unavailable were excluded.

Newborn Screening Procedure

Heel-prick dried blood spot (DBS) samples were collected on standardized filter paper between 24 and 72 hours after birth, according to the Saudi National Newborn Screening Program protocol [2,5]. Samples were allowed to dry completely before transportation to the National Health Laboratory under standardized conditions to ensure specimen integrity. Screening was performed using tandem mass spectrometry (MS/MS) for IEM disorders and immunofluorometric assays for endocrine disorders [5-9]. Positive screening results were communicated to the regional pediatric team for immediate clinical evaluation, confirmatory diagnostic testing, and initiation of appropriate management.

Disorders Included in the Screening Panel

- The national screening panel included 17 inherited metabolic and endocrine disorders, categorized as follows:
- Amino acid disorders: Phenylketonuria (PKU), Argininosuccinate Lyase Deficiency (ASLD), Maple Syrup Urine Disease (MSUD), and Citrullinemia (CIT).
- Organic acid disorders: Propionic Acidemia (PA), Methylmalonic Acidemia (MMA), Glutaric Acidemia Type 1 (GA-I), Isovaleric Acidemia (IVA), and 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC).
- Fatty acid oxidation disorders: Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) and Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD).
- Ketogenesis and ketolysis disorders: 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG) and Beta-Ketothiolase Deficiency (BKT).
- Carbohydrate disorder: Galactosemia (GALT).
- Endocrine disorders: Congenital Hypothyroidism (CH) and Congenital Adrenal Hyperplasia (CAH).
- Vitamin-responsive disorder: Biotinidase Deficiency (BTD).

Case Definition

National laboratories confirmed cases were included in the final analysis. Positive screening results were verified using confirmatory biochemical and/or molecular diagnostic testing in accordance with the Saudi National Newborn Screening Program guidelines.

Data Collection

Data were retrieved from the regional newborn screening database and included demographic information, screening coverage, confirmed diagnoses, and disease-

specific incidence. The incidence of each disorder was calculated as the number of confirmed cases divided by the total number of screened live births.

Statistical Analysis

Data was collected from newborn screening records for the study period of 1 January 2006 to 31 December 2019. Data were analyzed by using IBM SPSS Statistics (IBM Corp., Armonk, NY, USA). Descriptive statistics were used to summarize the study data. Categorical variables are presented as frequencies and percentage. Disease incidence was calculated by dividing the number of confirmed cases by total number of screened newborns (73,571) and is expressed as one per number of screened live birth (1:x). Exact 95% confidence intervals (CIs) were calculated for the overall incidence and major disorders using a Poisson distribution. Comparison with published national and incidence rate were descriptive and based on previously published reports.

Results

Screening Coverage

Between 1 January 2006 and 31 December 2019, a total of 75,600 live births were recorded in Ministry of Health hospitals in the Bisha region. Of these, 73,571 newborns (97.3%) underwent newborn screening through the Saudi National Newborn Screening Program. Among the screened newborns, 78 infants were confirmed to have inborn error of metabolic and endocrine disorders, corresponding to an overall incidence of approximately 1:943 live births (0.11%).

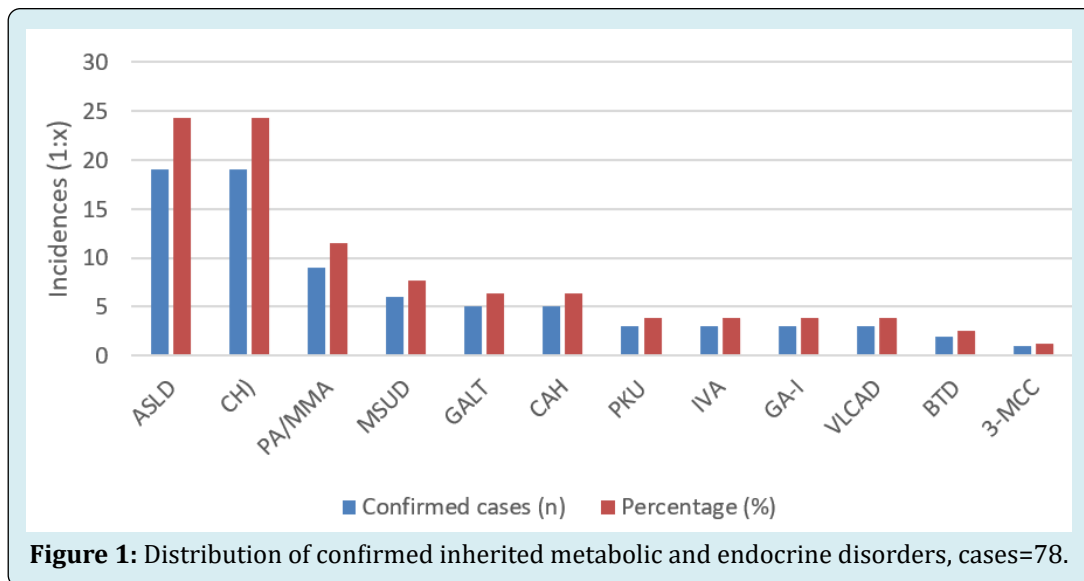
Spectrum of Detected Disorders

The distribution of confirmed disorders is presented in (Table 1, Figure 1) Argininosuccinate lyase deficiency (ASLD) was the most frequently identified disorder, accounting for 19 cases (24.36%), congenital hypothyroidism (CH) with 19 cases (24.36 %) and propionic acidemia/methylmalonic acidemia (PA/MMA) with 9 cases (11.54%). The remaining disorders included maple syrup urine disease (MSUD) (6 cases, 7.6%), galactosemia (GALT) (5 cases, 6.4%), congenital adrenal hyperplasia (CAH) (5 cases, 6.4%), phenylketonuria (PKU), isovaleric acidemia (IVA), glutaric acidemia type 1 (GA-I), and very long-chain acyl-CoA dehydrogenase deficiency (VLCAD), each with 3 cases (3.8%). Biotinidase deficiency (BTD) accounted for 2 cases (2.5%), whereas only one infant (1.28%) was diagnosed with 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC).

Disorder	Confirmed cases (n)	Percentage (%)	Incidence (1:x live births)
Argininosuccinate lyase deficiency (ASLD)	19	24.36	1: 3,872
Congenital hypothyroidism (CH)	19	24.36	1:3,872
Propionic acidemia/Methylmalonic acidemia (PA/MMA)	9	11.54	1: 8,174
Maple syrup urine disease (MSUD)	6	7.69	1: 12,262
Galactosemia (GALT)	5	6.4	1:14,714
Congenital adrenal hyperplasia (CAH)	5	6.4	1:14,714
Phenylketonuria (PKU)	3	3.85	1:24,523
Isovaleric acidemia (IVA)	3	3.85	1:24,523
Glutaric acidemia type 1 (GA-I)	3	3.85	1:24,523
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)	3	3.85	1:24,523
Biotinidase deficiency (BTD)	2	2.56	1:36,785
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)	1	1.28	1:73,571
Total	78	100	1: 943

Note: Incidence was calculated using 73,571 screened live births as the denominator .

Table 1: Distribution and incidence of confirmed inherited metabolic and endocrine disorders detected through the Saudi National Newborn Screening Program in the Bisha region, 2006–2019.



Disease Incidence

Disease-specific incidence rates are summarized in (Tables 1-6 & Figures 2-5). Among amino acid disorders, ASLD demonstrated the highest incidence (1: 3,872 live births), MSUD (1:12,262) and PKU (1:24,524). Within the organic acid disorders, PA/MMA showed the highest incidence (1:8,174 live births), whereas IVA and GA-I each occurred at

an incidence of 1:24,524, and 3-MCC was identified in only one newborn (1:73,571). Among endocrine disorders, CH was the most common (1:3,872), while CAH and GALT each demonstrated an incidence of 1:14,714. The incidence of BTD was 1:36,786. VLCAD deficiency was the only fatty acid oxidation disorder detected during the study period, with an incidence of 1:24,524.

Disorder	Bisha	Central Region	North/West Region	Eastern Region	Saudi Arabia	Global incidence
ASLD	1: 3,872	1:13,825	1:25,000	1:66,381	1:16,847	1:70,000
PKU	1:24,524	1:12,289	1:11,063	1:14,285	1:14,245	1:15,267
MSUD	1:12,262	1:22,120	1:4,224	1:4,285	1:15,816	1:83,333

Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Table 2: Comparison of the incidence of amino acid disorders between the Bisha region, other regions of Saudi Arabia, and international reports.

Disorder	Bisha	Central	Eastern	North/West	Saudi Arabia	Global
PA/MMA	1:8,174	1:12,289	1:25,000	1:39,828	1:14,090	1:93,457
IVA	1:24,524	1:11,060	1:25,000	1:24,892	1:29,807	1:196,078
GA-I	1:24,524	1:36,867	1:50,000	1:24,892	1:32,291	1:100,000
3-MCC	1:73,571	1:36,867	1:13,276	1:50,000	1:36,867	1:24,000–1:68,000

Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Table 3: Comparison of the incidence of organic acid disorders.

Disorder	Bisha	Central	North/West	Eastern	Saudi Arabia	Global
VLCAD	1:24,524	1:36,867	1:3,211	1:55,176	1:23,484	1:17,301

Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Table 4: Comparison of the incidence of fatty acid oxidation disorders.

Disorder	Bisha	Central	North/West	Eastern	Saudi Arabia	Global
CH	1:3,872	1:2,096	1:3,375	1:4,208	1:7,908	1:3,000–1:4,000
GALT	1:14,714	1:27,650	1:14,224	1:10,000	1:14,245	1:47,000
CAH	1:14,714	1:5,000	1:7,965	1:7,908	1:5,000–1:7,908	1:21,270
BTB	1:36,786	1:36,867	1:22,127	1:5,000	1:17,222	1:129,000

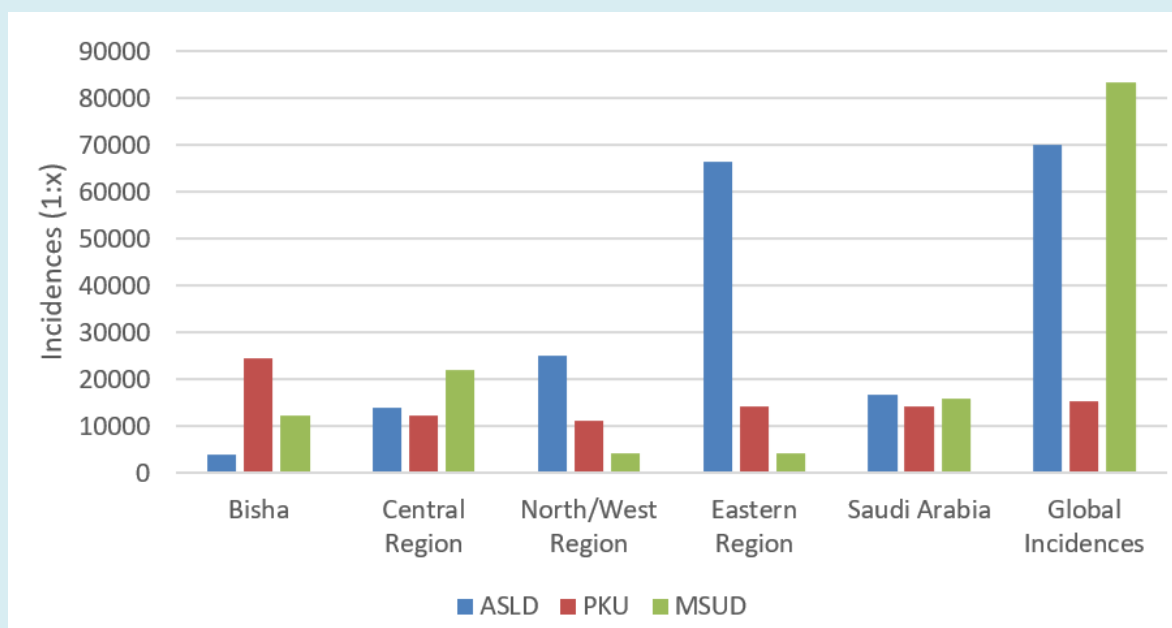
Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [12], Mak et al. [14] and Alratrout et al. [25].

Table 5: Comparison of the incidence of endocrine and vitamin-responsive disorders.

Bisha Region	1: 943
Regions of KSA	1:1,000 – 1:1,403
USA	1:4,000
Japan	1:7,000
Global	1:965-1:3,744

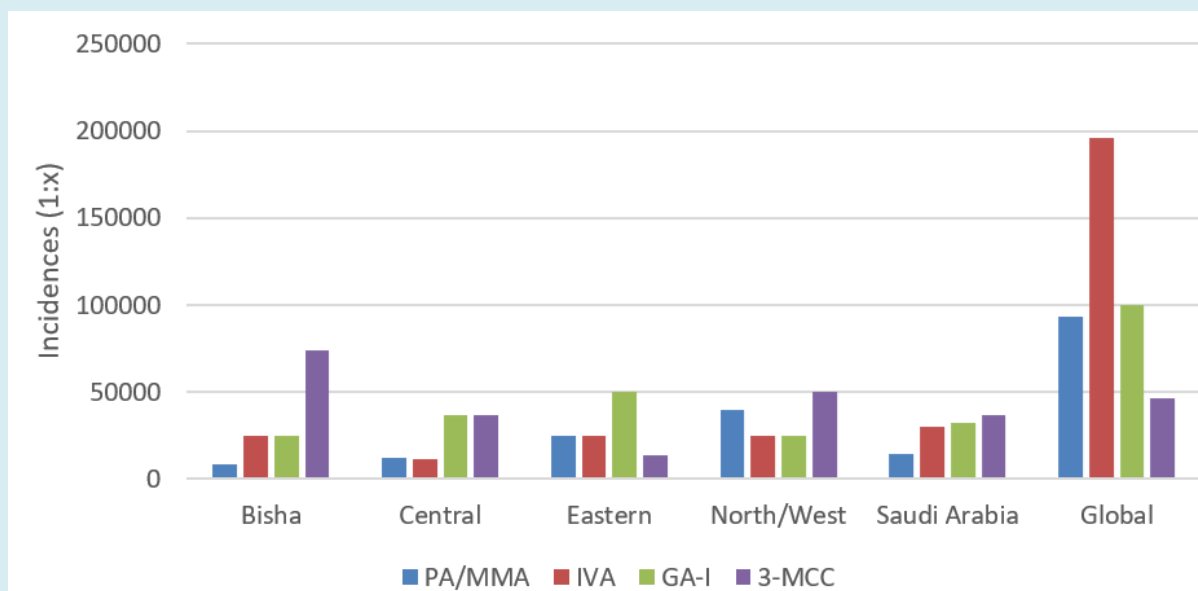
Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], and global incidences were derived from Waters et al. [15] and Mak et al. [16].

Table 6: Global comparison of overall Incidences of IEM disorders.



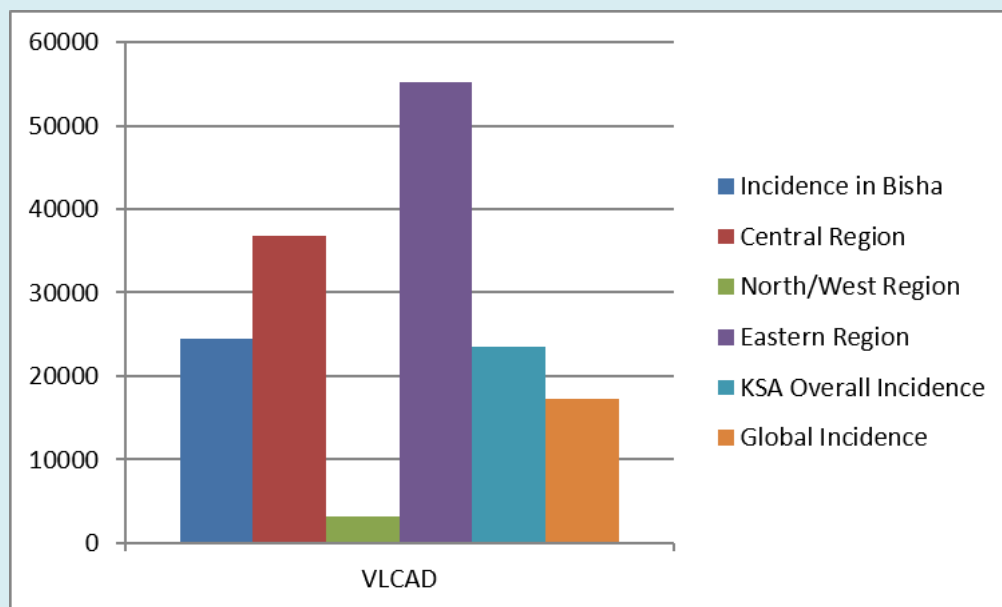
Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Figure 2: Comparison of aminoacidopathies incidence across regions.



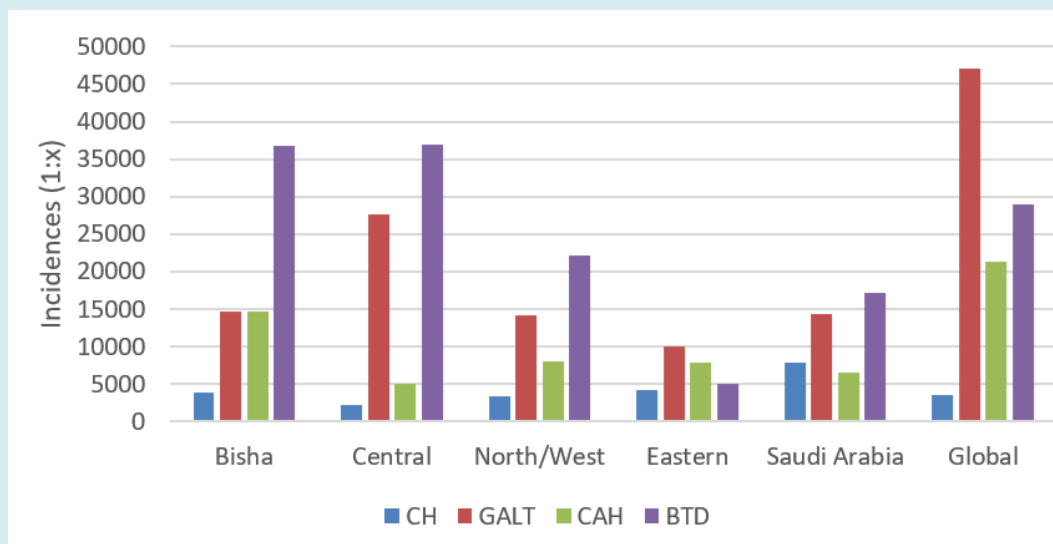
Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Figure 3: Comparison of organic acid disorders incidence across regions.



Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Figure 4: Comparison of VLCAD incidence across the regions.



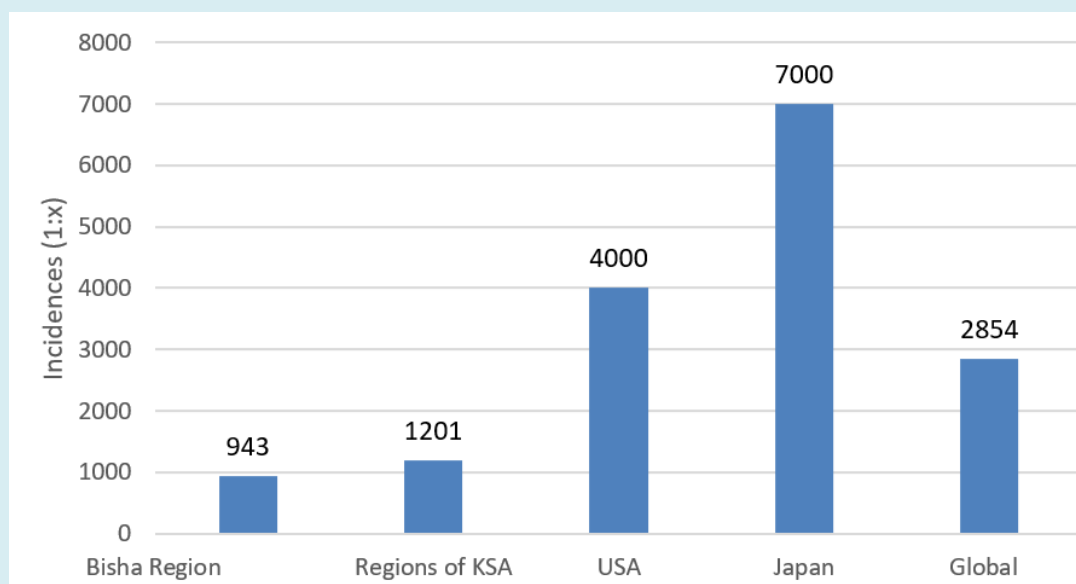
Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16] and Alratrout et al. [17].

Figure 5: Comparison of the incidence of endocrine and vitamin responsive disorders.

Comparison with Other Saudi Regions and International Data

The overall incidence of inborn error metabolism disorders in the Bisha region was high similar to what has been reported from some regions of Saudi Arabia. The most striking finding was the markedly increased incidence of ASLD, which were approximately four folds higher than that

reported in the Central Region and substantially higher than the national estimate. Likewise, the incidence of PA/MMA exceeded those reported from the Central, Eastern, and North/West regions. These comparative regional data were sourced from the national registry report by Alfadhel et al. [1], Alratrout et al. [17] while global incidence estimates were derived from international systematic reviews [15,16] (Table 6, Figure 6).



Note: Data for Saudi regional incidences were obtained from Alfadhel et al. [1], global incidences were derived from Waters et al. [15], Mak et al. [16].

Figure 6: Global comparison of overall incidences of IEM disorders.

Although congenital hypothyroidism remained the most common endocrine disorder nationally, its incidence in the Bisha region was comparable to that observed in other Saudi regions, whereas the incidences of PKU, CH, GALT, IVA and GA-1 were generally comparable to those reported in populations with established newborn screening programs (Tables 2-5 & Figures 2-5).

Discussion

This study presents the first comprehensive 14-year analysis of the Saudi National Newborn Screening (NBS) Program in the Bisha region and demonstrates a remarkably high incidence of inborn error of metabolism and endocrine disorders and higher incidence of ALS and PPA/MMA when compared with previously published national and international data. Among 73,571 screened newborns, 78 were confirmed to have one of the screened disorders, corresponding to an overall incidence of approximately 1 in 9,453 live births. This incidence considered high according to what has been reported from the Saudi National Newborn Screening Program [1,17] and is substantially higher than estimates from many Western and Asian populations [15-21].

The high incidence observed in Bisha is consistent with the well-recognized effect of consanguinity on the prevalence of autosomal recessive disorders in Saudi Arabia. Previous studies have shown that consanguineous marriage, particularly first-cousin marriage, remains common in many regions of the Kingdom and contributes significantly to the increased frequency of inborn error of metabolic and

endocrinal disorders [10,11], with particularly high rates of consanguinity documented in the Asir and Bisha provinces [12]. Founder variants within tribal populations may further increase the occurrence of specific disorders in certain geographic regions [10,11,16].

Argininosuccinate lyase deficiency (ASLD) was the most frequently detected inborn error metabolic disorder in the present study, with an incidence substantially higher than that reported from other Saudi regions and national estimates. Similarly, propionic acidemia/methylmalonic acidemia (PA/MMA) occurred at a higher frequency than national estimates. This regional clustering is likely driven by specific founder mutations within the local population, as previously documented for ASLD [21] and PA/MMA [22] in Saudi cohorts. These findings suggest regional genetic variation within Saudi Arabia and highlight the importance of reporting regional epidemiological data rather than relying solely on national averages.

Congenital hypothyroidism (CH) was the most common endocrine disorder identified, consistent with previous reports from Saudi Arabia and many international newborn screening programs. Early diagnosis through newborn screening allows prompt initiation of levothyroxine therapy, thereby preventing irreversible neurodevelopment impairment. Likewise, early identification of disorders such as PA/MMA, MSUD, VLCAD deficiency, and galactosemia enables dietary and medical interventions that substantially improve clinical outcomes and reduce mortality [2-7,19-24]. Although maple syrup urine disease (MSUD), galactosemia

(GALT), congenital adrenal hyperplasia (CAH), very long-chain acyl-CoA dehydrogenase deficiency (VLCAD), phenylketonuria (PKU), glutaric acidemia type 1 (GA-I), isovaleric acidemia (IVA), biotinidase deficiency (BTD), and 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC) were detected less frequently, their early identification remains clinically important because timely intervention can prevent life-threatening metabolic crises, irreversible neurological injury, severe developmental disability, and death. The identification of these rare disorders further demonstrates the effectiveness of tandem mass spectrometry-based newborn screening in detecting a broad spectrum of treatable inborn error of metabolic diseases before clinical presentation.

Compared with reports from Europe, North America, Japan, and other developed newborn screening programs, the incidence of inborn error of metabolic disorders in the Bisha region remains considerably higher [15-20], (Table 6, Figure 6), these differences are likely multifactorial and may reflect variations in population genetics, rates of consanguinity, founder effects, screening strategies, and case ascertainment. The widespread implementation of tandem mass spectrometry has also improved the detection of disorders that were previously under diagnosed.

The findings of this study support continued investment in the Saudi National Newborn Screening Program. In addition to universal newborn screening, strengthening genetic counseling, premarital screening, and targeted carrier testing for common regional disorders may reduce disease burden in high-risk populations [12,14,17,25]. Public health education aimed at increasing awareness of inherited disorders and reproductive risk should also be considered.

Strengths and Limitations

This study has several important strengths. It represents one of the longest regional newborn screening studies conducted in Saudi Arabia, covering a 14-year period with a high screening coverage rate (97.3%). All reported cases were confirmed through the Saudi National Newborn Screening Program, ensuring a high level of diagnostic accuracy. Furthermore, comparison with published national and international data provides valuable insight into regional variation in inborn error of metabolic disorders.

Several limitations should be acknowledged. First, the retrospective nature of the study limited the availability of detailed clinical and molecular genetic information for individual patients. Second, molecular confirmation and genotype-phenotype correlations were not available for all confirmed cases. Third, the study included only newborns delivered in Ministry of Health hospitals and may

not fully represent births occurring in private healthcare facilities. Finally, comparisons with other Saudi regions and international populations were descriptive and based on published data rather than direct statistical analyses.

Conclusion

Despite these limitations, this study provides important epidemiological evidence demonstrating that high overall incidence of 1:943 and a substantially higher incidence of ASLD and PA/MMA than what has been reported from other regions and internationally. These findings support continued investment in newborn screening, genetic services, and preventive public health strategies aimed at reducing the burden of inborn error of metabolism diseases. The results highlight the importance of maintaining universal newborn screening, strengthening genetic counseling services, expanding premarital and carrier screening programs, and enhancing public awareness of inborn error of metabolic disorders. Further multicenter studies incorporating molecular genetics analyses are recommended to improve understanding of the regional epidemiology and genetic basis of these disorders in Saudi Arabia.

Ethical Considerations

Approved by ethical committee of King Abdullah Hospital Bisha on 2021 and also no deal with human or animal tissue

Funding

The authors received no external funding for this study.

Conflict of Interest

No competing interest.

Data Availability

The data sets generated and analyzed during the current study are available from the corresponding author upon reasonable request.

Author contributions

Elwadiah Ziad: Conceptualization, study design, data collection, data interpretation, manuscript preparation.
Babatundi Kayode - Adedji: Statistical interpretation, critical revision, final approval of the manuscript
Nafiu Liadi: Data analysis, critical manuscript revision, literature review.

Footnotes

¹Incidence was calculated using 73,571 screened live births as the denominator.

²The overall incidence of IEM in Bisha region was high and a substantially higher incidence of ASLD and PA/MMA than what has been reported from other regions and internationally.

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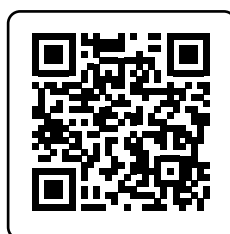
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