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EPIDEMIOLOGY OF INTELLECTUAL DISABILITY IN UPPER RURAL SWAT, KHYBER PAKHTUNKHWA, PAKISTAN

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Abstract

Background: Intellectual disability (ID) is a significant public health concern influenced by genetic, socioeconomic, and environmental factors. Epidemiological data from remote, high-consanguinity regions like Upper Swat, Pakistan, is limited. Objective: To investigate the epidemiology, demographics, and etiological factors of ID in Upper Rural Swat, Khyber Pakhtunkhwa, Pakistan. Methods: A cross-sectional study was conducted from December 2024 to May 2025 in Tehsil Bahrain. Data were collected from 209 clinically diagnosed participants using DSM-5 and ICD-11 criteria via snowball sampling. Statistical analysis was performed using SPSS v.27. Results: The highest prevalence was in the 10-19 years age group (38.8%). Moderate ID was most common (46.4%), followed by severe ID (30.6%). Males were significantly more affected (65.5%, $p<0.001$), and most cases were from rural areas (84.2%, $p<0.001$). Over half belonged to low-income families (54.5%, $p<0.001$). Parental consanguinity was a major risk factor, with 32.1% of cases from such unions; of these, 67.2% had a familial history of ID ($p<0.001$). Prenatal genetic disorders were the leading cause (62.2%). Conclusion: Intellectual disability in Upper Rural Swat is a substantial burden, strongly associated with consanguinity, low socioeconomic status, and rural residence. The findings underscore an urgent need for community-based genetic counseling, early screening, and targeted public health interventions.

INTRODUCTION

Intellectual disability (ID), also referred to as mental retardation (MR), is characterized by significant limitations in cognitive functioning and adaptive behavior, and it impacts an individual's capacity to live independently, especially when onset occurs at a young age (1). The World Health Organization (WHO) defines ID as significantly below-average cognitive functioning, at least two or more standard deviations below the mean, with a global prevalence estimated between 0.62% and 3.00% (2, 3, 4). This condition is heterogeneous; approximately 85% of cases are mild in nature (5). Individuals with ID experience challenges in three key areas: social, conceptual, and practical skills, which greatly affect their ability to navigate everyday life (6). Early signs of ID are varied and depend on factors such as age of onset, degree of impairment, and underlying biological causes, particularly in individuals with severe disabilities (7, 8). ID can often be identified in early childhood, and clinical diagnosis typically occurs within the first three years of life (9). Common symptoms include challenges with feeding or breathing, abnormal facial features, and delayed language development, which frequently lead families to seek medical care (10). As children enter school, academic difficulties and behavioral issues—such as attention deficits and mood swings—often become more pronounced, necessitating comprehensive clinical evaluation and personalized intervention (10). Other behavioral concerns, including self-harm and sleep problems, may also arise. Co-occurring psychiatric conditions like Attention-Deficit/Hyperactivity Disorder (ADHD), mood disorders, and anxiety are common, further complicating the clinical picture (7, 9, 11).

METHADOLOGY

This cross-sectional study investigated the prevalence and characteristics of intellectual disability (ID) in Tehsil Bahrain, Upper Swat, Khyber Pakhtunkhwa, Pakistan, from December 2024 to May 2025. Following ethical approval, participants were clinically diagnosed per DSM-5 and ICD-11 criteria, had a minimum one-year residency, and were of all ages except those above 70. Exclusion criteria included head injuries, specific learning disabilities, unrelated mental health conditions, recent migrants, non-consenting individuals, severe medical cases, and inaccessible remote residents. A snowball sampling technique, initiated through local educators and community elders, was used until data saturation. The assessment involved a clinical examination referencing the OMIM database, anthropometric measurements, and an evaluation of intellectual and adaptive functioning based on standardized diagnostic criteria. Data were analyzed in SPSS (v.27), using Chi-square tests to examine relationships between categorical variables, with statistical significance set at $p < 0.05$.

RESULTS

A detailed clinical and epidemiological assessment of 209 individuals with intellectual disability (ID) in Upper Rural Swat revealed significant trends across demographics, severity, socioeconomics, and etiology. The highest prevalence was observed in the 10-19 years age group (38.8%, n=81). Moderate ID was the most common severity level (46.4%, n=97), followed by severe (30.6%, n=64). A strong and significant male predominance was noted (65.5% vs. 34.4% female; $p < 0.001$), and the vast majority of cases resided in rural areas (84.2%, n=176; $p < 0.001$). Socioeconomic status was a critical factor, with over half of the affected individuals (54.5%, n=114) belonging to low-income families (earning < PKR 20,000), a disparity that was statistically significant ($p < 0.001$). The study found a powerful link between parental consanguinity and familial onset of ID. Among the 67 cases from consanguineous unions (32.1% of total), a striking 67.2% (n=45) had a familial history of ID, compared to only 23.9% (n=34) in non-consanguineous families ($p < 0.001$). Etiologically, prenatal factors were the leading cause, with genetic disorders accounting for 62.2% (n=130) of all cases. Among perinatal contributors, neonatal jaundice was notable (6.2%, n=13). Epilepsy emerged as the most frequent postnatal condition (18.2%, n=38). These findings collectively underscore a substantial burden of ID in this region, strongly associated with rurality, male gender, low socioeconomic status, and consanguineous marriage, with genetic prenatal factors constituting the primary etiological pathway

TABLE :*EPIDEMIOLOGY AND ASSOCIATED FACTORS OF INTELLECTUAL DISABILITY IN UPPER RURAL SWAT, PAKISTAN (N=209)*

CATEGORY / VARIABLE	SUB-CATEGORY	CASES (N)	PROP. (%)	95% CI / STATISTICAL DETAIL	P-VALUE
I. PREVALENCE BY AGE & SEVERITY					
AGE GROUP	<10 years	9	4.3%	(Mild:0%, Mod:4.1%, Sev:6.3%, Prof:5.6%)	N/A
	10-19 years	81	38.8%	(Mild:30.0%, Mod:43.3%, Sev:40.6%, Prof:22.2%)	
	20-29 years	62	29.7%	(Mild:40.0%, Mod:33.0%, Sev:21.9%, Prof:22.2%)	
	30-39 years	30	14.4%	(Mild:13.3%, Mod:12.4%, Sev:20.3%, Prof:5.6%)	
	40-49 years	18	8.6%	(Mild:10.0%, Mod:5.2%, Sev:7.8%, Prof:27.8%)	
	50-59 years	9	4.3%	(Mild:6.7%, Mod:2.1%, Sev:3.1%, Prof:16.7%)	
SEVERITY TOTALS	Mild	30	14.4%	Expected N: 52.3, Residual: -22.3	<0.001
	Moderate	97	46.4%	Expected N: 52.3, Residual: 44.7	
	Severe	64	30.6%	Expected N: 52.3, Residual: 11.7	
	Profound	18	8.6%	Expected N: 52.3, Residual: -33.3	
II. SOCIO-DEMOGRAPHICS					
GENDER	Male	137	65.5%	Expected N: 104.5, Residual: 32.5	<0.001
	Female	72	34.4%	Expected N: 104.5, Residual: -32.5	
LOCALITY	Rural	176	84.2%	Expected N: 69.7, Residual: 106.3	<0.001
	Semi-urban	29	13.9%	Expected N: 69.7, Residual: -40.7	
	Urban	4	1.9%	Expected N: 69.7, Residual: -65.7	
ECONOMIC STATUS (PKR)	Low (<20,000)	114	54.5%	Expected N: 52.3, Residual: 61.8	<0.001
	Low-Mid (20k-30k)	59	28.2%	Expected N: 52.3, Residual: 6.8	
	Mid (30k-60k)	28	13.3%	Expected N: 52.3, Residual: -24.2	
	High (60,000+)	8	3.8%	Expected N: 52.3, Residual: -44.2	
III. PARENTAL CONSANGUINITY					
MARRIAGE TYPE	Consanguinity	67	32.1%	Familial Onset: 67.2% (45/67)	<0.001
	No Consanguinity	142	67.9%	Sporadic Onset: 76.1% (108/142)	
IV. ETIOLOGICAL FACTORS				OMIM / ICD-11 Codes	
PRENATAL	Genetic (General)	130	62.2%	0.555 – 0.686	N/A
	Down Syndrome	8	3.8%	0.018 – 0.071 (OMIM:190685, ICD-11: LD40.0)	
	Environmental	4	1.9%	0.006 – 0.045	
PERINATAL	Neonatal Jaundice (NNJ)	13	6.2%	0.035 – 0.101 (ICD-11: KA82)	N/A
	Late Cry (Asphyxia)	5	2.4%	0.009 – 0.052 (ICD-11: KA81)	
	Cerebral Palsy	1	0.5%	0.001 – 0.022 (OMIM:603513, ICD-11:8A61)	
POSTNATAL	Epilepsy	38	18.2%	0.134 – 0.238 (OMIM:604233, ICD-11:8A60)	N/A
	Febrile Illness	9	4.3%	0.022 – 0.077 (ICD-11:MG26)	
	Low Dev. Milestones	1	0.5%	0.001 – 0.022	

CI = Confidence Interval; OMIM = Online Mendelian Inheritance in Man; ICD-11 = International Classification of Diseases 11th Revision. Statistical significance for Chi-square tests is set at $p < 0.05$.

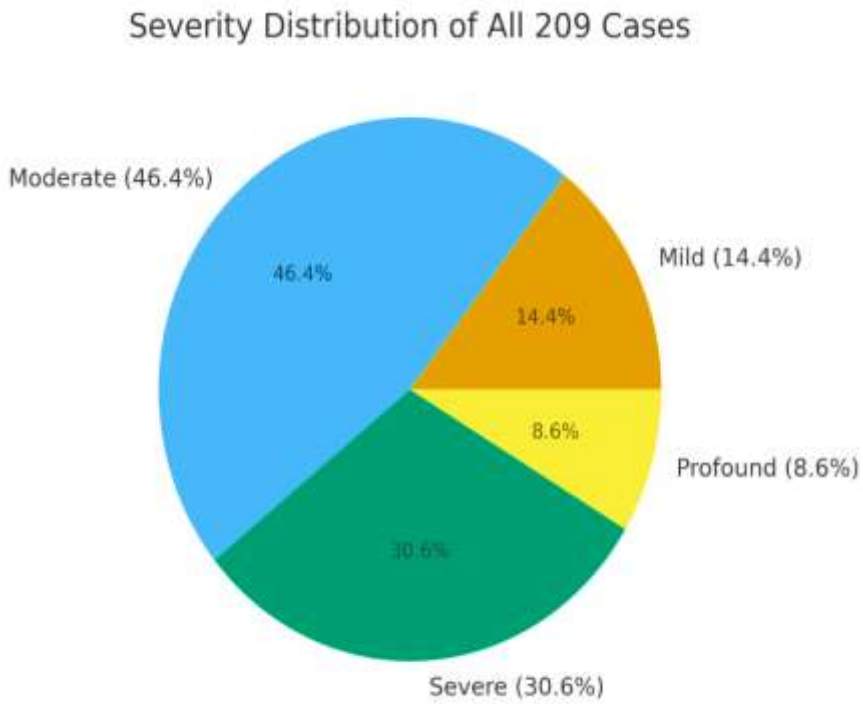

FIGURE 1. SEVERITY DISTRIBUTION OF ALL 209 CASES

DISCUSSION

This study conducts a comprehensive epidemiological and clinical analysis of intellectual disability (ID) within the distinctive social and cultural context of Upper Rural Swat. The region is characterized by closed family systems, frequent consanguineous marriages, and limited access to healthcare, all influencing health outcomes. Our findings indicate that ID among this population is multifactorial, arising from a complex interplay of genetic, environmental, and social determinants. The demographic distribution revealed a significant proportion of ID cases among adolescents and young adults, where the age group of 10–19 years accounted for 38.8% of cases, followed by those aged 20–29 years at 29.7%. This observation is consistent with global trends where ID often goes undiagnosed until school-aged or early adulthood, particularly in low- and middle-income countries (LMICs) like Pakistan [12, 13]. Previous studies have underscored variability in ID prevalence based on the selected population age groups, indicating the necessity for careful interpretation of epidemiological data [14, 15]. For instance, research from the Australian Bureau of Statistics suggested that the age-specific prevalence of disability is highest during childhood, peaking between 10 and 14 years and showing a decline in later adolescence [16]. The findings of this study showed a predominance of moderate (46.4%) and severe (30.6%) ID, aligning with research from other regions in Pakistan, where severe cases are more likely to be clinically recognized and reflected in survey data [17]. The lower frequency of mild ID cases (14.3%) in this study can be attributed to the snowball sampling technique, which may have led to underappreciation of less severe forms due to challenges in clinical and behavioral recognition. In examining gender distribution, the study found that males (n=137; 65.5%) were more frequently affected than females (n=72; 34.4%). This finding is congruent with literature reporting a higher prevalence of ID among males, potentially due to sex-linked vulnerabilities or diagnostic biases [18]. Additionally, the marked predominance of ID cases in rural households (n=176; 84.2%) and among low-income families (n=114; 54.5%) reflects trends observed in other contexts, where limited access to healthcare facilities and environmental factors exacerbate disability prevalence [19]. A pronounced association was noted between parental consanguinity and ID onset. Of the 209 cases examined, 32.1% occurred in consanguineous unions, with familial cases significantly more common in these families (67.2%) compared to non-consanguineous ones (23.9%). This aligns with research indicating that consanguinity is a prominent risk factor for neurodevelopmental disorders and congenital anomalies [20, 21]. The study also identified various prenatal, perinatal, and postnatal risk factors contributing to ID etiology. Genetic causes predominate, accounting for 62.2% of cases, with chromosomal abnormalities and genetic disorders

contributing substantially. Perinatal complications, such as neonatal jaundice (6.2%), and postnatal issues like epilepsy (18.2%), further inform the clinical picture of ID in this population. The associations observed are consistent with existing literature demonstrating the link between perinatal complications and later developmental outcomes [22].

CONCLUSION

This study highlights intellectual disability in Upper Rural Swat as a complex public health challenge driven by intertwined genetic, socio-economic, and environmental determinants. The high prevalence of moderate and severe ID, coupled with significant associations with parental consanguinity and low household income, emphasizes structural vulnerabilities within this rural population. Prenatal genetic causes dominated the etiological landscape, although preventable perinatal and postnatal complications also contributed meaningfully to disease burden. Rural residence and limited healthcare accessibility further amplified risks. Collectively, the findings underscore the need for multi-level interventions, including improved perinatal care, early developmental screening, community-based educational initiatives, and culturally appropriate genetic counseling programs. Addressing these factors can substantially reduce avoidable forms of ID and improve developmental outcomes in similar low-resource settings.

RECOMMENDATIONS

This study acknowledges several limitations, primarily the reliance on survey data that does not incorporate chromosomal or mutation analyses for definitive molecular diagnosis. Additionally, undetected physiological and metabolic abnormalities may exist. Factors affecting prenatal and postnatal mortality, maternal health, and pregnancy conditions require further exploration. Moving forward, it is essential to implement community-based prenatal screening, genetic counseling, and molecular diagnostic approaches to mitigate the risks associated with intellectual disability effectively.

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