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Bridging Genetics and Psychology: A Case-Based Exploration of Development in Noonan Syndrome

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Abstract

Noonan syndrome (NS) is a rare, autosomal dominant genetic disorder, which has an estimated occurrence of 1 in 1,000-2,500 live births. It is characterised by congenital heart defects, developmental delays and distinctive physical features, NS presents a multifaceted challenge in paediatric care. This case report focuses on the psychological and developmental presentation of a 3-year-old female from Mumbai diagnosed with NS. The main concerns were significant delays in motor and speech skills, as well as behavioural concerns such as inattention, sensory sensitivity, and difficulty in social communication. Considering the common comorbidity between NS and Autism Spectrum Disorders, the Indian Scale for Assessment of Autism (ISAA) was administered, indicating mild features of autism. Further assessments using the Developmental Screening Test (DST) and Vineland Social Maturity Scale (VSMS) suggested moderate developmental delays and mild socio-adaptive functioning. A comprehensive treatment plan was implemented, including speech therapy, occupational therapy, and pre-academic training over the course of six months. Post-intervention evaluations revealed significant improvements in socio-adaptive functioning and a decline in autism-related features, as reflected in improved scores on the same criteria. This case demonstrates the significance of early, multidisciplinary, and tailored interventions in improving outcomes for individuals with NS. The findings contribute to the limited literature on NS in the Indian population, encouraging for increased awareness, and early detection and care.

Keywords: Noonan Syndrome, Autism Spectrum Disorder, early intervention, case report

Introduction

Noonan Syndrome (NS), a heterogeneous genetic condition first identified by Noonan and Ehmke in 1963, is characterised by a variety of physical characteristics, congenital heart defects, developmental delays, and other abnormalities (McKusick, 1992; Noonan, 1968,

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1994). It occurs in approximately 1 in every 1000 to 2500 births, making it one of the most prevalent genetic syndromes after Down Syndrome (NIH, 2021).

NS is primarily caused by germline mutations in genes involved the RAS/MAPK (Rat sarcoma/mitogen-activated protein kinase) signalling pathway, which is essential for cell growth and development (Rauen, 2013). The most prevalent mutation, observed in about 50% of cases, occurs in the PTPN11 gene on chromosome 12q, resulting in an overactive SHP-2 protein and distinctive NS features (Tartaglia et al., 2001; Tartaglia et al., 2002). Additional mutations in genes such as SOS1 (Son of Sevenless Homolog 1), RAF1(RAF Proto-Oncogene Serine), KRAS (Kirsten Rat Sarcoma Viral Oncogene Homolog), NRAS (Neuroblastoma RAS Viral Oncogene Homolog), BRAF (B-Raf Proto-Oncogene), and RIT1 (Ras-Like Without CAAX 1) also contribute to NS's genetic diversity (Aoki et al., 2013).

Clinically, NS is characterised with distinct facial features, short stature, chest deformities, and cardiovascular abnormalities, particularly pulmonary valve stenosis and hypertrophic cardiomyopathy (Qazi et al., 1974). Developmental problems include delayed motor skills and intellectual abilities, with mild to borderline cognitive impairment typically observed (Pierpont et al., 2009). Speech and language impairments, attention deficits resembling ADHD, and social communication issues are also prevalent (Hopkins-Acos & Bunker, 2018; Lee et al., 2005).

Psychologically, individuals with NS may demonstrate anxiety, mood disorders, and difficulty with social integration, which are frequently associated with Autism Spectrum Disorder (ASD) (Wingbermühle et al., 2012). Learning difficulties impact visual-spatial processing and executive functioning, which influences academic performance (Pierpont, 2016). Despite these problems, many individuals with NS report satisfactory quality of life, highlighting the significance of tailored interventions and support (Sarimski, 2000; Shaw et al., 2007).

While substantial progress has been made in identifying genetic underpinnings of NS, Longitudinal investigations are required to better grasp long terms impacts and optimise therapeutic strategies (Pierpont et al., 2018). This case report aims to shed light on the psychological aspects of NS, with the goal to enhance holistic care strategies for affected individuals.

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Case Description

Index child, a 3-year-old girl, belonging to a middle socioeconomic, joint family from suburban Mumbai. At 3 years of age, the child was diagnosed with Noonan syndrome, a genetic disorder characterised by features of short stature, a receding hairline, low-set ears, curly hair, a broad forehead, hypertelorism (widely spaced eyes), dental anomalies, and ptosis (drooping of the upper eyelids).

With a continuous course and improving progress, the child presented with complaints chiefly characterised by developmental delays, delayed speech development, and behavioural disturbances. She presented with complaints of inattention, distractibility, low sitting tolerance, aggressive behaviour (head banging, biting, hitting others, and throwing objects), producing infantile squeals, difficulty adapting to change in routine, and exploring the surrounding by tactile and gustatory sensations (mouthing and licking of objects). Other concerns involve temper tantrums, mouthing, high screentime, disturbed sleep, and eating difficulties.

Client had one-word speech and communicated needs nonverbally through gestures. Socially, the child prefers being in groups but plays alone, and has few friends. Scholastic history revealed low average pre-academic training and discontinuing schooling after 6 months to receive specialised treatment for her developmental and behavioural issues.

There was no significant history suggestive of infectious disease, head injuries, and seizures. Family History revealed diabetes in maternal grandfather and hypertension in the paternal grandfather.

The child's prenatal history revealed an unplanned but wanted pregnancy, with both parents aged 27 at conception. The mother experienced gestational hypertension. Birth history revealed she is the only child, born preterm at 8 months due to premature rupture of membranes through caesarean delivery, with low birth weight (approximately 1.5 kgs). Neonatal history was significant for respiratory distress requiring supplemental oxygen, neonatal jaundice treated with two days of phototherapy and a month-long NICU stay due to a congenital heart defect and swollen kidney. The left kidney hydronephrosis was resolved by six months of age. Early cardiac evaluations detected a heart murmur, which is expected to resolve with age.

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The child was exclusively breastfed until four months, with semisolid foods introduced at six months. The developmental history shows severe delays in achieving age-appropriate milestones, particularly in crawling, sitting without support, speech, walking, and bowel and bladder control.

Behavioural observation revealed the child was well groomed and lean, appearing older than her age. Eye contact was made and maintained. She responded to her name on being called and presented a social smile. Attitude towards the examiner was cooperative. Working relationship was established. She was observed to smile to herself. She followed simple instructions. Attention was aroused but could not be sustained. Motor behaviour revealed rocking movements. Her temperament appeared to be slow-to-warm.

Treatment history revealed the child has been engaged in an early intervention focusing on speech therapy, occupational therapy, and special education. Primary focus included sensory integration, oral motor activities, eye-hand coordination, activities of daily living, supervised sitting, hand functionality with an emphasis on palmar grips, and command-based exercises. Emphasis was placed on gross motor skills, particularly trunk control and gait coordination, alongside pre-academic training eg. grip formation, sequencing, and social-communication development.

Pre- and post-assessments were administered in February and August, 2024, respectively, to monitor the developmental trajectory of the index child. Recent evaluations, conducted in August, 2024 of the revealed significant findings across multiple assessments. The Developmental Screening Test (DST) indicated a developmental quotient (DQ) of 43, indicating a moderate delay. The Vineland Social Maturity Scale (VSMS) showed a recent score of 60, reflecting a mild delay in socio-adaptive functioning, an improvement from the previous score of 52. Additionally, the Indian Scale for Assessment of Autism (ISAA) revealed a raw score of 61, indicating no features of autism spectrum disorder, marking a significant improvement from the previous score of 90, which suggested mild features of autism. An attempt was made to assess IQ using the Seguin Form Board Test (SFBT); however the administration was discontinued due to the child's inability to comprehend instructions and difficulty with shape recognition, preventing accurate task completion.

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Overall, the findings from psychological testing, behavioural observation, and interview information indicate mild to moderate delays in developmental and social-adaptive functioning with the primary diagnosis of Noonan syndrome.

Therapeutic interventions led to significant improvements in socio-adaptive functioning and a reduction in autistic features. Additionally, reductions in behavioural stimming, sensitivity to loud noises, and enhancements in socialisation, gait balance while climbing stairs, and group participation further underscore the treatment's effectiveness.

Discussion

Noonan Syndrome (NS) is a rare genetic condition that represents significant diagnostic barriers, particularly in India where its prevalence is not well explored. Because of limited awareness and the diverse clinical presentation of Noonan syndrome, several cases remain undiagnosed or become evident too late, postponing critical interventions (Vashishta et al., 2023; Dhillon & Fuks, 2021). In resource constrained settings like India, the detection of rare diseases like NS is especially challenging. Despite these challenges, early intervention is critical for addressing developmental delays and enhancing the quality of life for affected children (Romano et al., 2009; Tiemens et al., 2023).

In this case report, the index child demonstrated developmental impairments in motor and language skills which are important indications of NS (Pierpont et al., 2009). Early detection of these markers is crucial, since missed diagnoses can prevent interventions that drastically influence a child's developmental trajectory (Hopkins-Acos & Bunker, 2018).

To address these issues, a comprehensive early intervention plan was developed, incorporating a multidisciplinary approach that encompasses speech therapy, occupational therapy, sensory integration, and pre-academic training (Shaw et al., 2007; Aoki et al., 2013). This approach is critical for achieving optimal developmental outcomes and fulfilling the complex needs of children with NS.

The treatment plan addressed the child's developmental and psychological demands through structured goals. Immediate goals included developing therapeutic rapport through play-based therapy to encourage emotional expression and minimise anxiety, while regulating

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aggressive behaviours using behavioural interventions and increasing attention span with engaging activities.

Short-term goals included providing parental psychoeducation to enhance treatment adherence and foster a supportive environment, using speech therapy techniques such as picture cards and talking flashcards, for effective communication, and implimenting behavioural modification strategies to promote healthier coping mechanisms. Sensory integration therapy addressed sensory processing challenges, while physiotherapy improved gross motor skills.

Long-term goals included improving social skills through training and cognitive restructuring, creating individualised education plans (IEPs) for targeted academic support, and improving self-care abilities through occupational therapy. Continuous physiotherapy assisted the child's physical development, while emotional regulation techniques helped the child regulate his emotions. Regular developmental monitoring utilising assessments like the Developmental Screening Test (DST) and Vineland Social Maturity Scale (VSMS) assured the treatment plan remained effective and met with the child's evolving requirements.

The child's developmental progress showed a decline in Developmental Quotient (DQ) from 43 to 30.97, a finding consistent with the documented variability in developmental trajectories for children with NS (Pierpont et al., 2018). This variability highlights the importance of continuous monitoring and the flexibility to adapt therapeutic strategies as needed.

In conclusion, this case underscored the need for early, tailored interventions for children with Noonan syndrome (NS) to improve social-adaptive functioning and decrease maladaptive behaviours. Continuous monitoring and adaptable treatment plans are curucial due to the variable nature of NS.

Future research should prioritize on raising awareness, facilitating access to genetic services, and addressing systemic obstacles to diagnosis and treatment of rare genetic disorders in resource-constrained environments. Considering the comprehensive intervention plan, significant obstacles persist in management of NS in the healthcare system. Initiatives such as National Policy for Rare Diseases (2021) represent progress, yet persistent barriers, like insufficient funding, limited access to genetic services, and inadequate healthcare

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infrastructure, continue to impede the timely and accurate diagnosis of rare disorders such as NS (Tiemens et al., 2023).

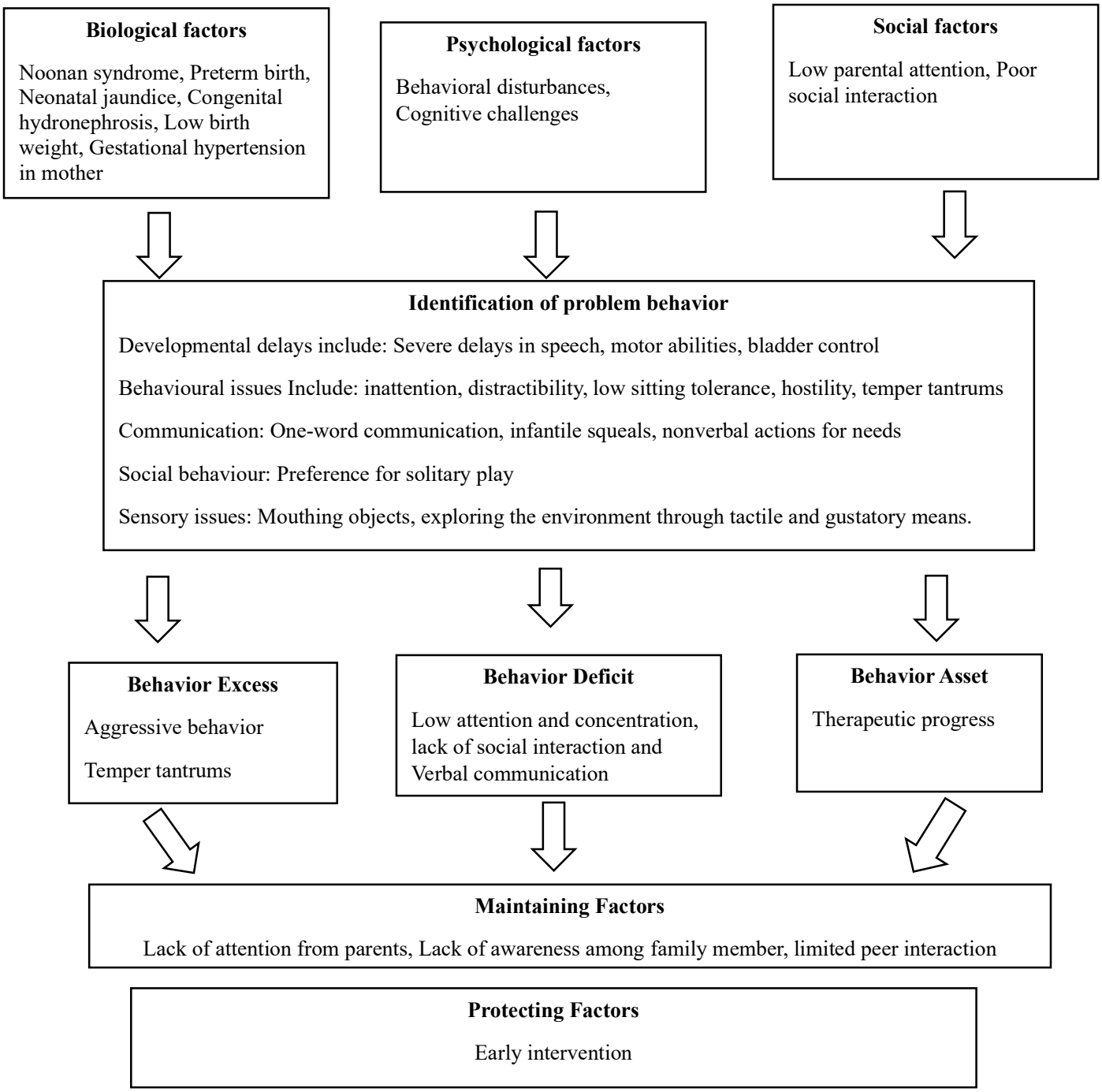

Fig 1. Case Conceptualisation

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