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MEETING PROCEEDINGS

NATIONAL CONFERENCE ON PRADER WILLI SYNDROME

24TH JANUARY 2026

BLiSC Campus, Bengaluru

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Note: The proceedings are from a scientific meeting. Please contact your healthcare provider for personalised medical advice.

About

Tata Institute for Genetics and Society (TIGS) along with Indian Prader Willi Syndrome Association (IPWSA) and Manipal Hospitals conducted National Conference on Prader Willi Syndrome (PWS) on 24th January 2026 at the Bangalore Life Science Cluster Campus. The aim of the meeting was to learn and discuss the multidisciplinary standard of care and recent developments in PWS management. The conference intends to nurture collaboration among PWS families, clinical experts like endocrinologists, nutritionists, pulmonologists, geneticists, researchers, allied health professionals working to improve the quality of life of individuals with PWS.

Understanding PWS

Prader-Willi Syndrome is caused by the loss of function of paternal genes in the SNRPN region and associated loci. Clinically, PWS presents with weak muscles at birth (hypotonia) and feeding difficulties in infancy, followed by excessive eating (hyperphagia) and obesity in later childhood, alongside hormonal, cognitive, and behavioural challenges. Like many rare genetic diseases, there is currently no cure for PWS, and effective care depends on coordinated, lifelong, multidisciplinary management.

Sleep Disorders in PWS

Dr. Ilin Kinimi (Consultant, Manipal hospitals, Bengaluru)

Dr. Illin Kinimi began her session by sharing her experience from 15 years ago while training in Singapore. She recalled how well the children with PWS were managed with regular sleep studies and BiPAP support, and upon returning to India, she noticed a significant gap in care standards. This realization motivated her to focus on building better sleep management resources specifically for the PWS community in India. She described the sleep issues progressive in nature and as a complex mix of factors like physical obstruction, hypothalamic dysfunction that affects sleep-wake regulation, leading to reduced sensitivity to drops in oxygen or spikes in carbon dioxide, meaning their bodies do not always react to danger signals of the oxygen drop during sleep which increases the risk for respiratory failure. Additionally, hypotonia and obesity can impact airway tone and passage eventually contributing towards obstructive sleep apnea, central sleep apnea, hypoventilation, excessive daytime sleepiness, in PWS. Dr. Kinimi urged parents to look beyond the obvious sign of snoring. She warned that silent problems are common and dangerous as child might not snore at all but could still be suffering from "quiet apnea" or shallow breathing called hypoventilation. She listed subtle red flags to watch for, such as morning headaches, restless sleep, sweating, or behavioural changes like irritability and poor attention in school. She also noted that these problems evolve with age, as detailed in the table below:

Age Group	Predominant Problems	Clinical symptoms
0-2 Years	Central control issues (brain forgets to breathe), Hypotonia.	Quiet apnea, cyanosis, poor arousal.
2-6 Years	Mixed pattern (Central + Obstructive), enlarged tonsils.	Snoring begins, restless sleep, morning irritability.
6-12 Years	Moderate to Severe Obstructive Sleep Apnea (OSA), obesity factors.	Loud snoring, witnessed apnea, learning issues.
Adolescence	Complex phenotype (Severe OSA + Hypoventilation), Narcolepsy-like features.	Sleep attacks, morning headaches, mood problems.

Dr. Ilin illustrated that these problems evolve in infants as they might struggle with their brain forgetting to breathe (central apnea), while older children often face obstruction due to enlarged tonsils or weight gain. On the topic of diagnosis, Dr. Kinimi was very firm about home sleep tests are not enough for PWS. She stated that a Level 1 in-lab Polysomnography with CO2 monitoring is the gold standard. She stressed that this testing is mandatory before starting Growth Hormone therapy with a follow-up test 6-12 weeks later, to ensure the hormone treatment induced growth of lymphoid tissues does not worsen the Obstructive Sleep Apnea. Growth Hormone therapy improves muscle tone, airway stability, reduces fat mass, however, can worsen OSA initially by lymphoid tissue growth and obstructing airway.

When discussing treatment, Dr. Kinimi also explained about non-invasive ventilation and strongly advocated for benefits of BiPAP machines over standard CPAP and how eventually children on CPAP might need BiPAP with severity of symptoms. She explained the difference simply, CPAP acts like a splint to keep the airway open and is beneficial for individuals with predominant OSA, BiPAP is advised for individuals who have OSA, hypoventilation and increased carbon dioxide and eventually BiPAP with a backup rate actually helps the child breathe if their brain pauses and majorly advised for central sleep apnea and hypoventilation with periodic follow ups. Dr. Kinimi also mentioned a critical safety warning regarding oxygen use, explaining that giving oxygen alone to a PWS child without ventilation support can be dangerous, it can cut off the respiratory drive causing the child to stop breathing and leading to a toxic buildup of carbon dioxide.

She concluded by acknowledging that while sleep equipment and studies can be expensive, they are a crucial investment in the child's development and safety. Dr Ilin remarked to be extremely cautious with anaesthesia, ensuring that any doctor performing a procedure is aware of the child's sleep risks and that monitoring is in place afterward. A multidisciplinary approach with pulmonology, endocrinology, ENT and sleep medicine is required for appropriate management of PWS.

Panel Discussion - Multidisciplinary Management of PWS

Manipal Hospitals, Bengaluru

Panelists

- Dr. Namratha Upadhya - Pediatric Endocrinologist
- Dr. Sham Kiran – Pediatric Endocrinologist
- Dr. Sushma Gopalan – Child Psychologist

Endocrine Management

1. Growth Hormone (GH) Therapy

Prader–Willi Syndrome is characterized by hypothalamic–pituitary dysfunction, resulting in near-universal growth hormone deficiency. Current consensus guidelines recommend initiating GH therapy between 3 and 6 months of age. With advances in genetic diagnostics, confirmation of PWS is now possible in the neonatal period, often within the first month of life, supporting earlier initiation of therapy.

Clinical Benefits:
GH therapy improves muscle tone and strength, optimizes body composition by reducing fat mass, enhances motor development, and contributes to improved cognitive outcomes. In infants, GH therapy significantly reduces the duration of early feeding difficulties associated with the initial nutritional phase.

Safety and Monitoring:

- Sleep Studies: A polysomnography is mandatory prior to initiating GH therapy in children older than 6 months, with a repeat study recommended 6–12 weeks after initiation. It is critical, as GH may increase adeno-tonsillar tissue and potentially worsen obstructive sleep apnea.
- Contraindications: GH therapy is contraindicated in individuals with severe obesity, uncontrolled severe obstructive sleep apnea, active malignancy, or active psychosis.
- Adverse Effects: Potential side effects include progression of scoliosis, disturbances in glucose metabolism, and intracranial hypertension, which may present as headaches.

GH therapy typically continued until completion of linear growth. However, in PWS, GH is prescribed beyond this period for metabolic benefits such as improved glycemic control and lipid regulation rather than height augmentation.

2. Thyroid Dysfunction

Approximately 20–30% of children with PWS exhibit thyroid abnormalities, most commonly central hypothyroidism. In this condition, impaired hypothalamic signaling results in inadequate stimulation of the thyroid gland. Few individuals with PWS are also known to have primary hypothyroidism and borderline low thyroid levels.

Screening and Diagnosis:
Diagnosis requires assessment of both free thyroxine (Free T4) and thyroid-stimulating hormone (TSH), as TSH levels may appear normal or low despite reduced T4 levels. Screening is recommended every six months and more frequently during initiation of GH therapy, as increased metabolic demand may unmask previously undetected hypothyroidism.

Management:
Treatment involves low-dose levothyroxine replacement, titrated according to biochemical parameters and clinical response.

3. Diabetes Mellitus

Individuals with Prader–Willi Syndrome are at increased risk of developing Type-2 diabetes mellitus due to obesity and insulin resistance, most commonly presenting during adolescence. Up to 40% of affected individuals may remain asymptomatic, lacking classic symptoms such as excessive thirst or urination. Routine screening using HbA1c is therefore essential. Treatment includes dietary modification, use of Metformin, and insulin therapy when required. GLP-1 receptor analogs have shown promise in adults but currently lack sufficient evidence for routine pediatric use.

4. Hypogonadism

Male infants frequently present with cryptorchidism and reduced penile length. Management may include human chorionic gonadotropin injections, although approximately half of affected children eventually require surgical correction. Pubertal development is often delayed, incomplete, or arrested. Hormone replacement therapy testosterone for males and estrogen for females is required not only for secondary sexual development but also for optimizing bone mineral density. In contrast, a subset of children may demonstrate features of early adrenal maturation, such as the pre-mature appearance of pubic hair.

Nutritional Aspects

1. Nutritional Phases

The infancy phase is marked by hypotonia and poor coordination of sucking, swallowing, and breathing, leading to feeding difficulties and failure to thrive. Strategies include waking lethargic infants for feeds, providing oral stimulation, and using specialized feeding bottles. When oral feeding is unsafe due to choking risk, tube feeding is required. GH therapy has been shown to shorten the duration of this challenging phase. A transition occurs during childhood when hyperphagia emerges, characterized by an intense drive to eat accompanied by impaired satiety. Due to reduced muscle mass and lower resting energy expenditure, children with PWS require approximately 60–80% fewer calories than their neurotypical peers to maintain a healthy body weight. Environmental control plays an important role; predictable meal and snack schedules reduce anxiety by providing clarity around food availability. Food should remain out of sight between meals, and grazing must be avoided. Uniform rules regarding food access should be maintained across home, school, and travel environment. Moreover, Food should never be used as a reward or punishment.

2. Dietary Composition

There is no single ideal diet for Prader–Willi Syndrome. Dietary plans should be balanced, rich in vegetables and fiber, and strictly portion controlled. Simple sugars should be avoided, as they can trigger intense cravings and exacerbate behavioral challenges.

Behavioral Aspects

1. Understanding Behavior

Behavioral challenges in Prader–Willi Syndrome arise from underlying hypothalamic dysfunction rather than intentional misconduct. Individuals often struggle with task switching and require structured flexibility. Sudden changes in routine can cause significant distress due to difficulty adjusting to expectations. Many individuals lack intrinsic mechanisms for regulating emotional responses.

2. Management Strategies

- **Co-Regulation**: During behavioral outbursts, caregivers must model calm behavior. Reasoning or negotiation during a meltdown is ineffective.
- **Safety First**: Ensuring safety and allowing space for de-escalation is essential. Once the individual is calm, returning promptly to the established routine prevents reinforcement of avoidance behaviors.
- **Visual Supports**: Visual schedules and advance transition warnings enhance predictability and reduce anxiety.
- **Play Therapy**: Early intervention through play therapy supports the development of neural pathways that reinforce adaptive behaviors.

A newly developed Prader–Willi Syndrome–specific behavioral screening tool created by Indian researchers enables systematic tracking of behavioral manifestations. This tool addresses a longstanding gap in condition-specific behavioral assessment and supports both clinical management and parental guidance.

Following the expert presentations and panel discussion, an in-depth and interactive question-and-answer session was conducted with active participation from parents, caregivers, clinicians, and healthcare professionals. The discussion focused on practical aspects of managing PWS, with particular emphasis on growth hormone therapy, feeding challenges, metabolic complications, respiratory support, puberty-related concerns, behavioural management, and long-term psychosocial issues.

Several parents sought clarification regarding IGF-1 levels and their role in determining eligibility for growth hormone (GH) therapy. Experts explained that in children with PWS, IGF-1 values should not be interpreted using absolute laboratory reference ranges alone. Instead, age- and sex-adjusted standard deviation scores are used to assess adequacy. Even if IGF-1 levels show numerical improvement over time, values falling below -2 SD (standard deviation) remain insufficient for the child's age and indicate benefit from GH therapy. It was emphasized that GH therapy is not initiated solely to improve linear growth, but also to support muscle tone, body composition, brain development, cognition, and overall neurodevelopment, particularly when started early in life.

Participants raised questions about the appropriate timing for initiating GH therapy, especially in infants with apparently normal height and weight. The panel clarified that normal growth parameters do not rule out the need for GH therapy in PWS. Current clinical practice favors early initiation once baseline assessments are completed, as early treatment has demonstrated long-term benefits beyond stature, including improved psychomotor outcomes. The concept of “why not start” rather than “why start” was emphasized, provided there are no contraindications.

Queries regarding daily v/s weekly GH injections were also discussed. Experts noted that weekly GH formulations have been recently introduced but are currently approved only for children with growth hormone deficiency unrelated to PWS. There is insufficient evidence supporting their use in PWS at present, and therefore daily GH injections remain the standard of care. Mixing daily and weekly regimens is not recommended until further research becomes available.

Concerns were expressed about temporarily stopping GH therapy during respiratory illnesses and the possibility of withdrawal effects. The panel clarified that GH should be withheld only during severe or critical illnesses, not during minor infections. Short interruptions do not reverse previously achieved benefits, and there is no recognized GH withdrawal syndrome. Episodes of excessive sleepiness during illness were explained as potentially related to post-infectious fatigue or central adrenal insufficiency, which can occur in a subset of individuals with PWS. Parents were advised to consider cortisol and ACTH evaluation during significant illnesses and to discuss stress-dose steroid coverage with their care team when indicated. Parents also enquired about baseline evaluations required before starting GH therapy. The experts outlined that assessment typically includes IGF-1 interpretation using standard deviation scores, thyroid function testing, documentation of baseline growth parameters, and a sleep study to assess for central or obstructive sleep apnea. Ongoing monitoring after initiation of therapy was stressed as an essential component of care.

Feeding concerns formed an important part of the discussion, particularly regarding poor weight gain, early introduction of solids, and obesity prevention. The panel emphasized that feeding decisions should be guided by developmental readiness rather than diagnosis alone. Indicators such as adequate head control, safe swallowing, and absence of choking are key determinants for introducing solids. If the child tolerates solids well, there is no need to discontinue them. Parents were reassured that obesity prevention is a

long-term goal and should not compromise appropriate nutrition in infancy. Support from physiotherapists and occupational therapists was encouraged to assist with feeding skills and safe progression. Questions were raised about breastfeeding in infants with poor sucking reflexes. Experts explained that sucking and breastfeeding skills are usually established early, and infants who have adapted to spoon feeding may or may not transition successfully to breastfeeding later. Gentle attempts may be made if feasible, but parents were reassured that inability to breastfeed should not be a source of concern. Emphasis was placed on introducing alternative feeding methods and supporting functional feeding skills through therapy.

Respiratory support **using BiPAP and associated behavioral resistance** was another area of concern. Parents described difficulties in initiating BiPAP use despite its clear benefits. The panel recommended **play-based desensitization strategies, family modeling, and creating positive associations** to reduce distress. Making respiratory support part of a routine and supportive environment was emphasized to minimize long-term aversion.

Puberty-related concerns in girls with PWS were addressed, including delayed puberty, menstrual irregularities, and the need for hormone replacement therapy. Experts noted that while many girls enter puberty spontaneously, progression may be delayed or incomplete. **Absence of breast development by around 13 years or lack of menstruation by 15 to 16 years requires evaluation**. Hormonal assessment, bone age studies, and pelvic imaging may guide decisions regarding estrogen replacement, which is generally affordable and may be required long-term for bone and cardiometabolic health. If menstrual cycles are regular, estrogen supplementation may not be necessary.

Diabetes and its interaction with GH therapy were extensively discussed. The panel explained that most diabetes in PWS is type 2 in nature and related to obesity and insulin sensitivity. Diabetes is not an absolute contraindication to GH therapy; however, poor glycemic control represents a relative contraindication. Optimization of blood glucose through diet, lifestyle modifications, and appropriate pharmacotherapy is essential before initiating or continuing GH therapy. Adults with PWS may still derive metabolic and musculoskeletal benefits from GH therapy at lower doses once diabetes is adequately controlled.

Behavioral issues and the timing of behavioral therapy were also explored. Experts explained that during the first two years of life, the focus remains on

physical health, development, and establishing structured yet flexible routines. Formal behavioral therapy is generally indicated later, only if challenges become difficult to manage or understand. Early establishment of predictable routines was highlighted as a protective factor against later behavioral difficulties. The challenges of **raising siblings alongside a child with PWS** were discussed, particularly in families with twins. Experts highlighted that families often undergo significant lifestyle restructuring, especially regarding food routines and household rules. Consistency across siblings helps prevent resentment and confusion. Open, age-appropriate communication fosters empathy and helps siblings develop a sense of shared responsibility and long-term support.

Finally, participants discussed disclosure of the PWS diagnosis to the child and the broader community. The panel emphasized that there is no fixed age for disclosure and that conversations should be gradual, age-appropriate, and rooted in trust. It was considered preferable for children to learn about their condition from family members rather than external sources. Decisions about disclosure to relatives, teachers, and schools should be phased and selective, taking into account the sensitivity and inclusiveness of the environment. Creating supportive, understanding communities was identified as a key determinant of long-term outcomes.

The session concluded with appreciation for the open dialogue between families and professionals, underscoring the importance of multidisciplinary care, early intervention, informed decision-making, and continued education in improving quality of life for individuals with Prader–Willi syndrome and their families.

Education, Employment & Insurance: Practical pathways

R. Anandhanayaki, Rehabilitation Officer, NIEPMD, Chennai

Ms. R Anandhanayaki, form the National Institute for Empowerment of People with Multiple Disabilities (NIEPMD) has two decades of experience in special education for persons with disabilities. Anandhanayaki introduced the concept of special education (SE) and history of how it started as a medical model and now has foraged to a technology-based model. Initially, SE was majorly about providing medical attention to people with disabilities. Later it moved to becoming a social model, wherein people were taught social and group behaviors transitioning to a vocational model and then to an educational model, which was all about providing basic education for individuals. Later, it took a leap and became an independent living model, where individuals were trained to live and perform all day-to-day tasks independently to live happy lives. After great efforts from many pioneers, it moved on to become a rights-based model, involving the legal and government authorities coming together and framing laws and rights that any PwD should be able to avail, this in true sense marked as a success in the sector. Currently, all these models are inculcated together with the help of technology, and Individual are taught to use and understand and take help of technology to thrive in this society. The speaker introduced The Rights of Persons with Disabilities (RPwD) Act, 2016, (RPwD Act 2016), enacted on December 27, 2016, and the SEPWD (Skill Council for Persons with Disability) had a major role to play in these developments.

Ms. Anandhanayaki mentioned the services and programmes offered by NIPEMD in the field of SE. Individualized Vocational educational programme & individual Family service programme, Skill Development Programmes, National Institute of Open Schooling – Vocational Programmes, HRD Programmes (in collaboration with Bharathidasan University, Trichy), Free coaching for persons with ID/ASD/SLD/MI & MD, Career Guidance & Placement Services, Linkage for Other Services & Benefits, etc.

Anandhanayaki shared the protocol that NIPEMD follows to train individuals describing a structured, rights-based pathway for vocational training and employment of persons with multiple disabilities, aligned with age-appropriate interventions. In early years (up to 14 years), the focus is on special schooling along with pre-vocational training to develop basic functional, social, and

work-readiness skills. During transition age (14–18 years), transition training is provided to prepare individuals for vocational environments and to identify suitable skill pathways. For adults (18 years and above), individualized vocational skill training is offered through independent living training, community-based training, simulated and on-the-job training based on ability and support needs. Appropriate employment options—open, supported, sheltered, or self-employment are then selected, followed by job placement, in-service training, and continuous follow-up to ensure job retention and sustainability, with support provided at both the institute and workplace in accordance with the RPwD Act, 2016 ensuring individuals can thrive in their job environment just like their colleagues.

RPwD Act, 2016 recognizes 21 types of disabilities grouped into broad categories. These include locomotor disabilities such as cerebral palsy, muscular dystrophy, dwarfism, leprosy cured, and acid attack victims; sensory disabilities covering visual impairment (blindness and low vision), hearing impairment (deaf and hard of hearing), and speech and language disability; intellectual disability, including specific learning disability and autism spectrum disorder; mental illness under mental and behavioral conditions; chronic neurological conditions such as multiple sclerosis and Parkinson's disease; blood disorders including hemophilia, thalassemia, and sickle cell disease; and multiple disabilities, which involve more than one of the listed conditions, including deaf blindness.

The speaker addressed the importance of family support and empowerment in making the individual and the family members live better. She explained that empowering the family is central to effectively supporting a person with disabilities. She spoke about the importance of first building awareness within the family about the individual's abilities, disabilities, and related conditions so that strengths are recognized and expectations are set realistic. She emphasized acceptance by parents, siblings, and extended family members, noting that emotional support at home directly influences the individual's growth and confidence. She then discussed the need to connect families with a wide range of resources, including financial assistance, medical and psychological care, therapeutic and training services, educational opportunities, and governmental, social, and legal support, with the family positioned as the core support system. She also highlighted advocacy, participation in support groups, and proper scheduling of activities as essential elements, stressing that a well-informed, engaged, and empowered family plays a crucial role in ensuring inclusion, stability, and long-term well-being.

Anandhanayaki explained the importance of inclusive education. Many universities are supporting inclusive education by allocating 5% reservations for PwDs which include the 21 disabilities listed in the RPwD Act, 2016. She talked about schemes like HEPSN and TEPSE that aid these individuals in higher education, by giving scholarships while improving accessibility and removing barriers. The legal frameworks like The United Nations Convention on the Rights of Persons with Disabilities UNCRPD 2006, National policy for persons with Disabilities 2006, RPwD Act 2016, The National Education Policy (NEP) 2020. She then proudly showed a list of all individuals she has trained and the jobs they are in.

She articulated that the challenges faced by people with disabilities evolve as they progress through different stages of education. Challenges are relatively fewer during the kindergarten and primary years, as the focus is mainly on early adjustment and basic learning. At the secondary level, challenges increase due to greater academic demands and social expectations. She further pointed out that inclusive education settings present a different set of challenges related to adaptation, accessibility, and peer interaction, while at the higher education level, challenges continue to develop as individuals work towards greater independence, advanced learning, and preparation for adult life and employment. Remarkable collaboration between Ability Foundation and Satyabhama University has led to successful completion of undergraduate and postgraduate programs for 50 individuals.

For adults with disabilities, the focus gradually shifts towards independent living and sustainable livelihoods, highlighting the wide range of skills required for everyday autonomy and work readiness. Developing employability skills alongside practical life skills such as travel training, money management, safety awareness, self-management, and community orientation, while also balancing home management, purchasing and banking skills, responsible use of the internet and gadgets, sex education, and overall life skills that support independent adult functioning. There are different types of employment available, including open and self-employment, supported and sheltered employment, cluster and parent–child supported models, as well as rural-based, voluntary, home-based, and seasonal employment, stressing that employment options must be flexible and individualized. Employment outcomes are closely linked to access to education, skilling and training opportunities, income generation, legal rights and support systems, and physical and social accessibility which overall constitute the key livelihood components .Selection

of appropriate employment depends on multiple factors such as the support of family members and friends, the individual's abilities and talents, community support, government and NGO assistance, targeted training in areas like money handling, stock management, social and communication skills, and exposure through internships and hands-on training, underscoring that successful employment is a collaborative and well-supported process. Career options should be explored from an early age of 15, which will enable appropriate planning and good execution. She ended her talk by providing a QR code link for all the government schemes and policies that can be helpful for individuals and their families. Later discussions involved questions about PWS not falling under any of 21 disabilities mentioned in the RPwD Act 2016, to which the speaker answered saying, individuals with PWS can be considered in the Intellectual disability category.

Saujanya Tata (Nayi Disha)

Nayi Disha has helped close to 9 Lakh family caregivers. Nayi Disha is a parent-centric organization that has social workers and counsellors helping families with information, guidelines, and protocols to avail various government and non-government facilities. She noted the importance of an individual with a disability to have a Unique disability Identity (UDID) card, which can help in accessing all the facilities provided by central and state governments. This is implemented by the Department of Empowerment of Persons with Disabilities to build an integrated system for the issuance of Universal ID and Disability certificates. This card includes demographic details and disability-related information of individuals across the country. The documents required to avail an UDID card are original Aadhaar card of the individual with disability, a passport size picture, and signature or thumb impression. In some cases, an old disability certificate might also be required. The application for UDID card can be found on swavalambancard.gov.in website. Nayi Disha portal is a mobile-first multilingual platform that guides parents through the process of availing different government schemes, multimedia resources for families, and a national directory of disability service providers. They also have multiple parent support communities that enable communication and emotional support from other families, thus empowering families in a community of families.

The speaker enlisted the various schemes and benefits available for people with disabilities, focusing mainly on education-related support and financial assistance. There are educational provisions where at least 5% of seats in

government and semi-government institutions are reserved for people with disabilities, upper age limits are relaxed by five years, and students are entitled to accommodations such as extra exam time, scribe or writers, grace marks, and proper seating arrangements. Saujanya highlighted the role of National Institute of Open Schooling (NIOS), where students are given up to five years to complete their studies and appear for examinations, and noted that inclusive education under the RTE Act is supported through schemes like Sarva Shiksha Abhiyan. She then spoke about need-based provisions under NIOS, explaining that parents must apply with a disability certificate at the time of exam registration to access facilities like additional time, short breaks during exams, scribes, assistive devices, and computer use. Moving on to scholarships, both pre-matric for classes 9 and 10, and post-matric from class 11 onwards, covering maintenance allowances, book allowances, disability allowances, and in post-matric cases, partial fee reimbursement, with applications can be done online through the national scholarship portal. These schemes aim to reduce barriers and ensure equal access to education and support services for people with disabilities.

She then went on to explain the health insurance, assistive device support, travel concessions, and tax benefits available for people with disabilities in detail. She described the Niramaya Health Insurance Scheme, pointing out that it is meant for people with cerebral palsy, intellectual disability, autism, and multiple disabilities, with coverage up to ₹1 lakh per year on a reimbursement basis and no age or income limit, provided claims are submitted within 30 days and the scheme is renewed annually. She briefly outlined what the insurance covers, including hospitalization, OPD care, therapies, alternative medicine, dental care, and transportation costs, all within specified sub-limits. She then spoke about the Assistance to Disabled Persons for Purchase/Fitting of Aids/Appliances (ADIP) scheme, explaining that it supports people with disabilities by providing free or subsidized assistive devices, prosthetics, educational kits, and learning aids, with full assistance for those earning below ₹22,500 per month and partial assistance for those earning slightly above this limit, and mentioned the documents required for application through implementing agencies.

Regarding travel-related benefits, she explained that persons with disabilities are eligible for railway and airfare concessions depending on the type and percentage of disability, along with facilities like wheelchair assistance, priority check-in, and escort concessions, and she also detailed the process and documents required to apply for an Indian Railway Concession Card. Finally, she spoke about income tax benefits, explaining the deductions available under

Sections 80DD for caregivers and 80U for persons with disabilities, highlighting the higher deductions for severe disability and briefly guiding how to claim these benefits online with the required medical certification and UDID details. Ms. Shika also mentioned that she has initiated conversations with a private insurance company providing health insurance for individuals with Prader Willi syndrome, which if successful would be a great accomplishment by IPWSA.

The session concluded by mentioning the helpline number of Nayi Disha team which can guide parents through many legal and social policies that are made for the benefit of individuals and their families. Helpline number: 084484 48996 (available 24*7).

Genetics of PWS

Dr. Gayatri Iyer, TIGS, Bengaluru

Dr. Gayatri Iyer illustrated the fundamental concepts of genetics including DNA, inheritance and central dogma of molecular biology. She explained what genes are and discussed their role in determining different health conditions, disease susceptibility, and drug metabolism. There are about 200 genes in humans wherein only one parental copy of gene is active and other is silenced by DNA methylation. As the aberration is with DNA methylation, genetic tests like sequencing or chromosomal microarray cannot appropriately diagnose imprinting disorders. Certain assisted reproductive methods like In Vitro Fertilization (IVF) can potentially increase the incidence of imprinting defects. Different aberrations on chromosome 15 can lead to two starkly different conditions of Prader Willi or Angelman syndromes depending on parent of origin. In PWS, the paternal complement of chromosome 15q11-13 region undergoes aberrations like deletion (60-70%), Uniparental Disomy (UPD) (30-40%), and imprinting defects (2-5%). Loss of paternal copy of genes lead to clinical characteristics of endocrine dysfunction, metabolic concerns and cognitive challenges.

Dr. Gayatri described the different genetic changes and their recurrence risk in subsequent pregnancies. Most cases of PWS are de novo, indicating, they were novel in the individual and the recurrence risk is less than 1% to the siblings. In upto 3% of individuals, there could be chromosomal rearrangements or one of the parents could carry genetic changes which can increase the risk of recurrence in subsequent cases. It is important to undergo genetic counselling along with genetic testing to understand the recurrence risk and plan the prenatal diagnosis for subsequent pregnancy wherever indicated. The type of genetic changes can be an early indication of the spectrum of clinical features that can be observed and contribute towards the differences in clinical profiles of individuals with PWS. Current diagnostic approaches used for PWS include Methylation-Specific Multiplex Ligation-dependent Probe Amplification (MS-MLPA) and Methylation specific polymerase chain reaction (MS-PCR). Prenatal testing such as amniocentesis strictly should be followed by MS-MLPA for definitive diagnosis.

Dr. Gayatri also spoke about the work that is carried out by her team in Tata Institute for Genetics and Society (TIGS) with key focus on study of Imprinting

Disorders (IDs). The team currently is developing a simple, rapid and cost-effective MS-PCR for PWS, Angelman Syndrome, Beckwith Wiedemann and Silver Russell syndromes which is intended to be used as first line testing strategy in any basic molecular lab with a simple PCR machine. They are also developing panels for imprinting disorders using long read sequencing technology, with a focus on 13 different disorders and pharmacogenomics. Pharmacogenomics is study about associated genes and their interaction with prescribed drugs. Different individuals can respond differently to the same drug based on their genetic makeup. She spoke in detail about prescription management in PWS.

Dr. Iyer briefly outlined the study protocol being followed by her team which integrates cost effective diagnostic strategies. Majority of the literature is from western population which does not provide accurate characteristics of PWS cohort from India. Dr. Gayatri invited all delegates to participate in an online survey about the clinical spectrum of PWS individuals observed in India which can reveal unique characteristics and challenges faced by individuals here. She concluded her speech by expressing gratitude to her team members, collaborators, funding organization, and parent advocacy groups for their continued support.

Living with PWS

Maahi Saxena

Maahi Saxena is an aspiring psychologist, dancer, and swimmer. She shared her educational and personal journey with the audience, beginning with her school life, where she spoke about her experiences with bullying. She articulated how she was often an easy target due to weight-related stigma and being fat shamed from a very young age. She articulated how it took her considerable amount of time and effort to build confidence to stand where she is today, highlighting the importance of self-belief and resilience. During her speech, she referenced a quote from Mahabharata which was told by Lord Krishna to Radha, which served as a source of strength and motivation throughout her life. Mahi is currently pursuing a Bachelor of Arts (Honours) degree from Indira Gandhi National Open University (IGNOU). She spoke about the life changing powers of counsellors in her life and expressed heartfelt gratitude to the doctors who supported her and family members who played a pivotal role in her life. She shared the challenges related to food habits in individuals with PWS, highlighting difficulties associated with managing food cravings. She shared her personal strategies for coping with these challenges by creating a structured routine and disciplined plans. She spoke about her interests which included chess and swimming, learning dancing from YouTube videos, paper quilling, playing the piano etc. Swimming is an important part of her life. Maahi expressed her determination and courage to rise again whenever she was faced with any setbacks. The session concluded with a video montage of her life inspiring the audience with her journey.

Concluding Remarks

The summit ended with closing remarks from Ms. Shikha Metharamani, the president of IPWSA sharing the journey of the team and way forward. Shikha reminisced how the group has come a long way and how awareness and management of rare genetic disorders has evolved in India over the years. She mentioned the future milestones IPWSA has set for itself like establishing medical insurance for individuals with PWS and enabling experts to develop generic/biosimilars for approved medications in India to enable accessibility and improve quality of life of individuals with PWS.

We invite individuals living with Prader–Willi Syndrome (PWS) and their caregivers to participate in a research study conducted by TIGS to help advance understanding of PWS in the Indian context.

Scan the above QRcode to fill the form.

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