GUIDING LIGHTS: EMPOWERING DOWN SYNDROME FAMILIES

STUDY PROTOCOL:

Protocol Metadata & Research Team

- **Study Title:** Guiding Lights: Empowering Down Syndrome Families
- **Research Framework:** Comprehensive Nutritional, Clinical, Developmental, and Psycho-Educational Investigation
- **Principal Investigator:** Zakira Asharef (Diploma in Nutrition and Health Education [DNHE], IGNOU; Department of Clinical Psychology, Manipal College of Health Professions [MCHP], MAHE)
- **Co-Investigators & Academic Collaborators:** Anila Hari (Dr. Anila H. L., Assistant Professor), Zahra Asharef, Aswin Koshy
- **Collaborating Institution:** Happy Child Special Needs School "Down Syndrome" Rehabilitation Project, Seiyun, Hadramout, Yemen
- **Target Population:** Children and adolescents diagnosed with Down Syndrome (Trisomy 21) aged 2–18 years and their families/caregivers
- **Registry Identification:** ISRCTN Registry (Submission ID: 50339)

Executive Summary

This study protocol outlines the operational framework for "Guiding Lights: Empowering Down Syndrome Families," an interdisciplinary research and community-oriented intervention project in Seiyun, Hadramout, Yemen. Children and adolescents with Down Syndrome face multifaceted physiological, metabolic, nutritional, and developmental vulnerabilities, while their families frequently encounter marked caregiving stress and social barriers.

Rather than relying strictly on standard deficit-oriented metrics or static IQ scores, this protocol implements a holistic, family-centered approach. We systematically evaluate anthropometric growth, micronutrient and metabolic markers, daily dietary habits, and functional milestone achievement, connecting these assessments directly to structured parental education and practical support. Conducted in partnership with the Happy Child Special Needs School, the protocol defines feasible methodologies and targeted interventions across three age cohorts (2–5 years, 6–11 years, and 12–18 years) tailored for low-resource settings.

Background and Rationale

Clinical and Biological Context

Down Syndrome (Trisomy 21) is the most prevalent chromosomal condition associated with intellectual disability, affecting approximately 1 in 700 live births globally. The presence of an extra copy of chromosome 21 alters gene expression across multiple organ systems, producing characteristic craniofacial features, generalized musculoskeletal hypotonia, immunological alterations, and neurodevelopmental variations.

Children with Down Syndrome present with a high prevalence of co-occurring medical conditions that require ongoing clinical vigilance:

- **Congenital Heart Defects (CHD):** Present in 40% to 60% of cases, commonly including atrioventricular septal defects (AVSD) and ventricular septal defects (VSD).
- **Endocrine Dysregulation:** High susceptibility to thyroid dysfunction, starting as subclinical hypothyroidism in early childhood and often progressing to overt autoimmune hypothyroidism.
- **Gastrointestinal and Oral-Motor Challenges:** High rates of reflux, chronic constipation, and feeding difficulties caused by oral-motor hypotonia.
- **Nutritional and Micronutrient Vulnerabilities:** Heightened risk for vitamin D deficiency, compromised iron stores (ferritin < 15 ng/mL), secondary microcytic anemia, and atypical metabolic shifts predisposing to central adiposity as children age.

Developmental and Functional Trajectory

While most individuals with Down Syndrome function within mild to moderate cognitive delay ranges, static IQ scores rarely reflect daily potential. Children frequently show strong visual learning capabilities, high socio-emotional perception, and receptive language skills that markedly surpass their spoken language abilities. When supported with consistent nutritional and educational guidance, children frequently achieve daily functional competencies well beyond what static IQ scores predict.

Psychosocial Context in Low-Resource Settings

In regions such as Seiyun, Yemen, access to specialized developmental care, pediatric subspecialties, and structured therapy is severely constrained. Families frequently navigate diagnosis and care with minimal formal guidance, leading to caregiver stress and isolation. At the same time, parents demonstrate deep dedication and resilience. Providing caregivers with clear health literacy, practical nutritional advice, and peer support networks transforms caregiver distress into proactive family empowerment.

Theoretical Model: The Four Pillars

Pillar	Focus Area	Key Objectives & Interventions
Pillar 1	Nutritional Support & Guidance	Age-tailored dietary guidance, texture-modified feeding strategies for hypotonia, macro/micronutrient adequacy, and proactive obesity prevention.
Pillar 2	Health Literacy & Monitoring	Routine surveillance for thyroid disorders, growth monitoring using Down syndrome-specific standards, sensory screening, and micronutrient tracking.
Pillar 3	Parental Awareness & Peer Networks	Practical caregiving workshops, structured psycho-education, stress-reduction strategies, and local caregiver support circles.
Pillar 4	Developmental Milestone Tracking	Multi-domain functional evaluations to guide home stimulation and support individualized educational planning (IEP).

Study Objectives

Primary Aim

To establish, implement, and evaluate an integrated clinical, nutritional, and psycho-educational protocol ("Guiding Lights") that assesses the physiological and functional status of children with Down Syndrome while delivering family-centered guidance in a community setting.

Specific Objectives

- **Growth Profiling:** Evaluate physical growth parameters across developmental stages using standard anthropometry and Down syndrome-specific growth references.
- **Dietary Assessment:** Quantify caloric intake, dietary diversity, and texture adaptations using repeated 24-hour recalls and Food Frequency Questionnaires (FFQ).
- **Biochemical & Clinical Surveillance:** Track key biomarkers including complete blood counts, thyroid profiles (TSH, free T4), serum ferritin, and 25(OH) vitamin D.

- **Functional Milestone Evaluation:** Profile cognitive, motor, linguistic, and adaptive milestones using structured developmental screening scales.
- **Caregiver Needs Assessment:** Document parental stress, coping mechanisms, and practical educational priorities.
- **Implementation:** Translate findings into personalized nutritional plans and actionable home developmental goals for each participating family.

Study Design and Technical Workflow

Study Setting & Cohort Stratification

- **Setting:** Happy Child Special Needs School, Seiyun, Hadramout, Yemen.
- **Target Sample Size:** n = 53 children and adolescents with cytogenetically or clinically confirmed Trisomy 21 and their primary caregivers.
- **Sampling Rationale:** A complete-enumeration sample of all registered, eligible students attending the rehabilitation center.

Cohort	Developmental Stage	Age Range	Primary Focus
Cohort 1	Early Childhood	2–5 Years	Early motor milestones, oral-motor feeding, thyroid and micronutrient status.
Cohort 2	Primary School	6–11 Years	School readiness, speech development, balanced nutrition, weight regulation.
Cohort 3	Adolescence	12–18 Years	Functional life skills, metabolic health, adolescent autonomy, peer socialization.

Eligibility Criteria

- **Inclusion Criteria:** Confirmed diagnosis of Down Syndrome (Trisomy 21); aged between 2 and 18 years; registered at or receiving services through the Happy Child Special Needs School network; written informed consent provided by a parent or legal guardian.
- **Exclusion Criteria:** Presence of acute, unstable medical emergencies requiring immediate inpatient hospitalization; primary caregiver unable or unwilling to complete assessment sessions.

Data Collection Tools & Procedures

Anthropometric Assessment

Physical measurements are conducted by trained examiners following standard anthropometric protocols:

- **Body Weight:** Measured to the nearest 0.1 kg using calibrated electronic digital scales.
- **Length / Height:** Recumbent length (for children under 2 or unable to stand) using an infantometer, and standing height measured to the nearest 0.1 cm using a vertical stadiometer.
- **Body Mass Index (BMI):** Calculated as weight (kg) divided by height squared (m^2).
- **Mid-Upper Arm Circumference (MUAC):** Measured at the midpoint between the acromion and olecranon using a flexible, non-stretch tape to evaluate peripheral muscle mass and acute undernutrition.

Nutritional & Biochemical Surveillance

- **Dietary Intake:** Repeated 24-hour dietary recalls administered to primary caregivers combined with a regionalized Food Frequency Questionnaire (FFQ) reflecting local Yemeni dietary staples.
- **Biochemical Markers:** Fasting blood samples collected at affiliated local laboratory facilities: Complete Blood Count (CBC), serum ferritin, 25-hydroxyvitamin D, Thyroid Stimulating Hormone (TSH), and free T4.

Developmental & Functional Rating Scales

- **ABCD Developmental Screening Form:** Assesses functional ability across four key domains: (A) Cognitive skills, (B) Gross and fine motor coordination, (C) Communication and language, and (D) Social-emotional behavior.
- **Multidimensional Educational Rating Scale:** Evaluates daily life skills and learning independence on a 5-point scale:
    - **1 = Not Yet Observed:** No independent demonstration.
    - **2 = Emerging:** Observed only with direct physical or verbal prompts.
    - **3 = Occasionally Demonstrates:** Shows intermittent independence in structured settings.
    - **4 = Frequently Demonstrates:** Consistent execution requiring minimal cues.
    - **5 = Mastered:** Fully independent across natural home and school environments.

Study Timeline & Implementation Phases

- **Phase 1: Institutional Approvals & Tool Adaptation (Months 1–2):** Finalizing institutional permissions, translating assessment questionnaires into Arabic, and conducting local pilot testing with educators.
- **Phase 2: Baseline Clinical & Nutritional Profiling (Months 3–5):** Enrolling participants, obtaining consent, collecting anthropometric measurements, dietary recalls, and laboratory samples.

- **Phase 3: Developmental Assessments & Family Workshops (Months 6–9):** Conducting ABCD milestone evaluations, administering caregiver questionnaires, and running Four Pillars parent education sessions.
- **Phase 4: Data Synthesis & Community Dissemination (Months 10–12):** Completing quantitative/qualitative data analysis, preparing individualized parent summary reports, and submitting documentation for registry updating and peer-reviewed publication.

Data Management and Statistical Analysis

- **Quantitative Analysis:** Continuous variables (anthropometry, laboratory parameters) will be summarized as mean ± standard deviation (SD) or medians with interquartile ranges. Categorical data will be expressed as counts and percentages. Comparative analyses across age cohorts will utilize independent t-tests, one-way ANOVA, or non-parametric alternatives (Mann-Whitney U, Kruskal-Wallis) where distributions depart from normality.
- **Qualitative Synthesis:** Caregiver survey responses and open-ended feedback will undergo thematic analysis to identify common barriers to healthcare and schooling.
- **Data Security:** All hard-copy evaluation records are stored in secure archives at the partner facility. Electronic data is stripped of direct personal identifiers, labeled with unique participant codes (e.g., GL-01 to GL-53), and stored on encrypted, password-protected drives accessible only to authorized study investigators.

Ethical Governance & Declarations

- **Ethics & Consent:** The protocol operates in accordance with the Declaration of Helsinki. Written informed consent is obtained from all primary caregivers prior to enrollment, alongside child assent wherever feasible.
- **Clinical Referral Mechanism:** Immediate referral protocols are established with local healthcare providers if severe medical concerns (e.g., untreated severe thyroid deficiency, severe acute anemia, or cardiac symptoms) are identified during screening.
- **Trial Registration:** Registered with the ISRCTN Registry under study identifier 50339.
- **Funding & Competing Interests:** Conducted as part of academic research requirements under the DNHE curriculum (IGNOU) in academic collaboration with MCHP, MAHE. The authors declare no competing financial or personal conflicts of interest.
- **Limitations:** The study utilizes a non-randomized, institutional cohort of 53 children within Seiyun, which may limit generalizability to rural populations outside structured educational programs. Ongoing regional infrastructure constraints also limit access to advanced pediatric subspecialty diagnostics.

References & Methodological Guidelines

American Academy of Pediatrics. (2022). Health Supervision for Children and Adolescents With Down Syndrome. *Pediatrics*, 149(5), e2022057010.

World Health Organization. (2006). *WHO Child Growth Standards: Length/height-for-age, weight-for-age, weight-for-length, weight-for-height and body mass index-for-age*. World Health Organization.

Smith, E., et al. (2018). Parental stress, coping strategies, and life satisfaction among caregivers of children with developmental disabilities. *Journal of Applied Research in Intellectual Disabilities*, 31(2), 220–231.

National Down Syndrome Society (NDSS). *Comprehensive Resources for Healthcare and Development in Down Syndrome*.

Individuals with Disabilities Education Act (IDEA), Part C: Early Intervention Program for Infants and Toddlers with Disabilities.

Appendices: Data Collection Instruments

Appendix A: Down Syndrome Clinical & Nutritional Assessment Form (ABCD Survey)

(Adapted for field screening at Happy Child Special Needs School)

- **Section 1: General Information & Demographics:** Child's full name, DOB, gender, parent/guardian details, contact and residence info.
- **Section 2: Anthropometric Data:** Height (cm), weight (kg), head circumference, BMI, arm span, waist/hip circumference, skinfold thickness (triceps, subscapular).
- **Section 3: Biochemical & Laboratory Parameters:**
    - Complete Blood Count (CBC): RBC, Hb/Hct, total/differential WBC, platelets, red cell indices (MCV, MCH, MCHC, RDW).
    - Thyroid Function Profile: TSH, Free/Total T3, Free/Total T4, Thyroid autoantibodies (TPOAb, TgAb, TSI, Thyroglobulin).
    - Glycemic & Lipid Profiling: Fasting glucose, OGTT, HbA1c; Total cholesterol, LDL, HDL, triglycerides, non-HDL.
    - Hepatic & Renal/Electrolytes: ALT, AST, ALP, GGT, direct/total bilirubin, albumin, total protein, urinalysis, serum electrolytes (Na, K, Cl, HCO3).
    - Micronutrient Status: 25-Hydroxyvitamin D, serum ferritin, serum iron, TIBC, serum calcium.
- **Section 4: Clinical Symptoms & Organ Surveillance:** Overall health rating, congenital heart defect monitoring, respiratory symptoms and sleep apnea signs, gastrointestinal function/motility (constipation/reflux), musculoskeletal hypotonia, neurological history (seizures, delays), sensory screening (vision/hearing).
- **Section 5: Dietary Profile & Feeding Behaviors:** Habitual meal patterns, fruit/vegetable intake frequency, protein/carbohydrate/fat sources, fluid balance, dietary supplementation, feeding difficulties/oral-motor fatigue, allergies/food intolerances.
- **Section 6: Informed Consent & Assent Block:** Formal caregiver authorization and signature.

Appendix B: Down Syndrome Child & Family Psycho-Educational Survey Form

(Parent/Caregiver Instrument)

- **Section 1: Basic Demographics:** Grade level, primary home language, age, gender.

- **Section 2: Medical History & Service Utilization:** Co-occurring medical diagnoses, healthcare visit frequency, current pharmacotherapy.
- **Section 3: Developmental Milestones & Interventions:** Age at gross motor and expressive milestones (sitting, crawling, walking, first words); therapies currently accessed (Speech, Occupational, Physical, Behavioral).
- **Section 4: Education & Learning Trajectory:** Educational setting (special education vs. mainstream/homeschool), classroom support structure (IEP, special education classroom, dedicated aide), parent-rated academic progress.
- **Section 5: Social & Emotional Well-Being:** Peer interaction patterns, extracurricular participation, emotional distress/anxiety manifestations.
- **Section 6: Parental & Caregiver Support Dynamics:** Caregiver self-efficacy rating, active support services (respite care, parent circles, workshops), identified gaps and needed family support resources.
- **Section 7: Open-Ended Caregiver Qualitative Feedback:** Qualitative remarks on systemic barriers and specialized needs.

Appendix C:استمارة مسح الأطفال المصابين بمتلازمة داون

استمارة مسح الأطفال المصابين بمتلازمة داون

القسم 1: المعلومات الأساسية

1. اسم الطفل: _____
2. العمر: _____
3. الجنس:
   - ☐ ذكر
   - ☐ أنثى
   - ☐ أخرى: _____
4. مستوى الصف الحالي: _____
5. اللغة الأساسية المستخدمة في المنزل: _____

القسم الثاني: التاريخ الطبي

1. هل يعاني طفلك من أي حالات طبية أخرى تم تشخيصها؟ (مثل أمراض القلب واضطرابات الغدة الدرقية)
   - ☐ نعم
   - ☐ لا
   - إذا كانت الإجابة بنعم، يرجى التحديد: _____
2. كم مرة يقوم طفلك بزيارة أخصائي الرعاية الصحية؟
   - ☐ شهريا
   - ☐ ربع سنوي
   - ☐ نصف سنوية
   - ☐ سنويا
   - ☐ أخرى: _____
3. قم بإدراج أي أدوية يتناولها طفلك حاليًا: _____

القسم 3: معالم النمو

1. في أي عمر حقق طفلك المعالم التالية؟ (يرجى التقريب)

- الجلوس بشكل مستقل: _____

- الزحف: _____

- المشي: _____

- الكلمات الأولى: _____

2. هل يتلقى طفلك أيًا من العلاجات التالية؟ (حدد كل ما ينطبق)

- ☐ علاج النطق
- ☐ العلاج الوظيفي
- ☐ العلاج الطبيعي
- ☐ العلاج السلوكي
- ☐ أخرى: _____

القسم 4: التعليم والتعلم

1. ما نوع البيئة التعليمية التي يعيش فيها طفلك حاليًا؟

- ☐ المدرسة السائدة
- ☐ مدرسة التربية الخاصة
- ☐ التعليم المنزلي
- ☐ أخرى: _____

2. ما نوع الدعم التعليمي الذي يتلقاه طفلك في المدرسة؟ (حدد كل ما ينطبق)

- ☐ برنامج التعليم الفردي (IEP)
- ☐ فصل التربية الخاصة
- ☐ مساعد/مساعد المعلم
- ☐ أخرى: _____

3. كيف تقيم التقدم الأكاديمي لطفلك؟

2. ما هي أنواع خدمات الدعم التي تستخدمها حاليًا؟ (حدد كل ما ينطبق)

- ☐ مجموعات دعم أولياء الأمور
- ☐ الاستشارة/ العلاج
- ☐ الرعاية المؤقتة
- ☐ ورش عمل تعليمية
- ☐ أخرى: _____

3. ما هي خدمات الدعم الإضافية التي قد تكون مفيدة لك؟ _____

القسم 7: التعليقات الإضافية

يرجى تقديم أي تعليقات أو معلومات إضافية تعتقد أنها ستساعدنا على فهم احتياجات طفلك ودعمها بشكل أفضل: _____

ملاحظة: تم تصميم نموذج الاستبيان هذا لجمع معلومات عن احتياجات وتجارب ورفاهية الأطفال المصابين بمتلازمة داون. يُستخدم نموذج الاستطلاع هذا للحصول على معلومات من أولياء الأمور أو مقدمي الرعاية أو المتخصصين الذين يعملون مع هؤلاء الأطفال.

Appendix D: القسم 1: معلومات عامة

القسم 1: معلومات عامة

1. الاسم الكامل للطفل:
2. تاريخ الميلاد:
3. الجنس:
4. اسم ولي الأمر/الوصي:
5. معلومات الاتصال:

- رقم التليفون:
- عنوان البريد الإلكتروني:
- عنوان المنزل:

القسم 2: بيانات القياسات البشرية

1. الارتفاع (سم):
2. الوزن (كجم):
3. محيط الرأس (سم):
4. مؤشر كتلة الجسم (BMI):
5. امتداد الذراع (سم):
6. محيط الخصر (سم):
7. محيط الورك (سم):
8. سماكة ثنيات الجلد (العضلة ثلاثية الرؤوس، تحت الكتف، إلخ. بالملم):

Section 3: Biochemical Parameters

1. Blood Tests:

- *Complete Blood Count (CBC) (attach results)*
  RBC:
  Hgb or Hb:

Hct:
WBC:
WBC Differential
Neutrophils:
Lymphocytes:
Monocytes:
Eosinophils:
Basophils:
Platelet Count:
MCV:
MCH:
MCHC:
RDW:

- *Thyroid Function Tests (attach results)*
  TSH:
  Free T3:
  Free T4:
  Total T3:
  Total T4:
  Thyroid antibodies
  TPOAb:
  TgAb:
  TSI :
  Thyroglobulin:

- *Blood Glucose Levels (attach results)*
  Fasting Blood Glucose Test:
  Oral Glucose Tolerance Test (OGTT):
  Random Blood Glucose Test:
  HbA1c (Hemoglobin A1c) Test:

- *Lipid Profile (attach results)*

Total Cholesterol:
LDL:
HDL:
Triglycerides:
Non-HDL Cholesterol:

- *Liver Function Tests (attach results)*
  ALT:
  AST:
  ALP:
  CGT:
  Total Bilirubin:
  Direct (Conjugated) Bilirubin:
  Albumin:
  Total Protein:

2. Vitamin and Mineral Levels:

- *Vitamin D (attach results)*
  25-Hydroxyvitamin D Test:

- *Iron/Ferritin (attach results)*
  Ferritin Levels:
  Serum Iron:
  TIBC:

- *Calcium (attach results):*

- *Others (specify and attach results):*

3. Other Relevant Tests:

- *Urinalysis (attach results):*

- *Electrolytes (attach results)*
  Sodium level:
  Potassium level:
  Chloride level:
  Bicarbonate level:

القسم 4: الأعراض السريرية

1. **الصحة العامة:**

- الحالة الصحية العامة: (ممتاز، جيد، مقبول، ضعيف)
- أي أمراض أو دخول إلى المستشفى مؤخرًا: (نعم/لا، قم بالوصف)

2. **أعراض القلب:**

- أمراض القلب (مثل عيوب القلب الخلقية):
- الأعراض (على سبيل المثال، ألم في الصدر، وضيق في التنفس):

3. أعراض الجهاز التنفسي:

- مشاكل في الجهاز التنفسي (مثل الالتهابات المتكررة، وانقطاع التنفس أثناء النوم):
- الأعراض (مثل السعال والأزيز):

4. أعراض الجهاز الهضمي:

- مشاكل في الجهاز الهضمي (مثل الإمساك والارتجاع):
- الأعراض (مثل آلام البطن والانتفاخ):

5. أعراض الجهاز العضلي الهيكلي:

- آلام المفاصل أو العضلات: (نعم/لا، قم بالوصف)
- نقص التوتر العضلي (ضعف العضلات): (نعم/لا)

6. الأعراض العصبية:

- النوبات: (نعم/لا، التكرار)

- تأخر النمو: (وصف)

7. أعراض أخرى:

- ضعف السمع أو الرؤية: (نعم/لا، قم بالوصف)
- الحالات الجلدية: (مثل الأكزيما والصدفية)

القسم 5: الملف الغذائي

1. العادات الغذائية:

أ) النظام الغذائي اليومي النموذجي: (وصف الوجبات والوجبات الخفيفة)

ب) تفضيلات الطعام: (الأطعمة المفضلة، الأشياء التي لا تحبها)

ج) أي أنظمة غذائية خاصة مُتبعة: (على سبيل المثال، خالية من الغلوتين، خالية من الكازين)

2. المدخول الغذائي :

أ) تكرار تناول الفواكه والخضروات: (يوميًا، أسبوعيًا، نادرًا)

ب) مصادر البروتين: (مثل اللحوم ومنتجات الألبان والنباتات)

ج) مصادر الكربوهيدرات: (مثل الحبوب والحبوب)

د) مصادر الدهون: (مثل الزيوت والزبدة والمكسرات)

3. تناول السوائل:

أ) كمية المياه اليومية (لتر):

ب) المشروبات الأخرى المستهلكة (مثل العصير والصودا):

4. المكملات الغذائية:

أ) الفيتامينات أو المكملات المعدنية: (نعم / لا، القائمة)

ب) المكملات العشبية أو البديلة: (نعم / لا، القائمة)

5. سلوك التغذية والأكل:

أ) الشهية: (جيدة، عادلة، فقيرة)

ب) أي صعوبات أو قيود في التغذية: (وصف)

6. الحساسية الغذائية أو عدم تحملها:

أ) الحساسية المعروفة: (نعم / لا، القائمة)

ب) أعراض الحساسية أو عدم التحمل: (وصف)

القسم 6: الموافقة والاتفاق

أوافق على التقييم وجمع البيانات:

● أوافق على تقييم واستخدام المعلومات المقدمة في هذا النموذج لفرض دعم نمو طفلي. (نعم/لا)

● توقيع ولي الأمر/الوصي:

● التاريخ:

** تعليمات تعبئة النموذج **

- يرجى ملء كل قسم بأفضل ما تستطيع.

- إذا كان السؤال غير قابل للتطبيق، فيرجى الإشارة إلى ذلك بـ "غير متاح".

- إرفاق أي تقارير طبية أو نتائج فحوصات ذات صلة.

- لأية تعليقات أو استفسارات إضافية، يرجى استخدام المساحة المتوفرة في نهاية كل قسم أو إرفاق صفحات إضافية إذا لزم الأمر.