Diagnosis in neurodevelopmental disorders: time, dynamics, and response to intervention

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The Tension

- **Current Practice:** Diagnostic reasoning relies heavily on **cross-sectional observations** (a single time point).
- **The Conflict:** Early intervention is expected, yet diagnostic validity depends on how difficulties **evolve over time**.
- **The Missing Link:** Limited integration of "time" into diagnostic models.

Key Concepts:

- **Dynamic Manifestations:** Symptoms are not static; they emerge from brain maturation, environmental demands, and compensatory processes.
- **Heterogeneity of Trajectories:** Especially in childhood, maturational differences can lead to divergent interpretations of the *same* behaviors.
- **Illustrative Marker:** Executive Functions (prolonged maturation, transdiagnostic nature, sensitivity to contextual demands).
- **Paradigm Shift:** Diagnosis is not a one-time decision, but a **process grounded in developmental trajectories**.
- **Clinical Implication**: Longitudinal observation is critical to distinguish **transient** difficulties from **persistent** neurodevelopmental constraints
- **Take-Home Message):** *In neurodevelopment, diagnosis is not the reading of a state, but the interpretation of a trajectory."*

Introduction:

- **The Public Health Burden:**
- **NDDs:** High prevalence, long-term impact on cognition, education, and social function.
- **Systemic Pressure:** Access to support is tied to a formal diagnosis.
- **The Clinical Reality:** Clinicians must make categorical diagnoses when stability and future course are **uncertain**.
- **The Epidemiological Paradox (ADHD as an Example):**
- **Lifetime Diagnosis Rates:** ~15.5% of US adolescents (12-17).
- **Current Prevalence:** Substantially lower (~9-11%).
- **Interpretation:** Highlights the influence of **developmental dynamics** on diagnostic outcomes.
- **Key Insight:** Diagnosis is not a fixed state; it fluctuates over time.

Challengs

- **Absence of validated biomarkers**
- **Phenotypic Heterogeneity**
- **Dynamic Context:** Manifestations emerge from brain maturation, environmental demands, and compensatory processes.
- *How should "developmental time" be integrated into clinical decision-making?*
- **The Aim of This Perspective:**
- To examine how developmental time informs diagnostic reasoning.
- **Illustrative Marker:** Executive Functions (prolonged maturation, transdiagnostic, context-sensitive)

2.The Diagnostic Paradox & Early Intervention

- **1. The Promise of Early Intervention:**
  - **Objective:** Earlier recognition → Earlier support → Potential to influence developmental trajectories.
  - **Evidence:** Demonstrated benefits in language, social, and academic domains (Dawson et al., 2010).
  - **Priority:** Early identification is a priority in clinical practice, public health, and education.
- **2. The Mismatch in Clinical Reasoning:**
  - **Expectation:** Parents, teachers, and primary care providers expect **rapid diagnostic categorization**.
  - **The Medical Model:** Inherited from other fields where identifying a disorder leads directly to classification.
  - **The Limitation:** In neurodevelopment, this logic is limited by the **absence of validated biomarkers**
- **3. The Diagnostic Paradox**.

Conclusion:

Developmental trajectories and response to intervention are **not equally integrated** into diagnostic reasoning across disorders, settings, and clinicians

1.Time as a Core Dimension:

- Time is not just a background variable; it is a **central dimension of assessment**.

2. Trajectories Under Intervention:

- Observing changes under intervention reveals distinct trajectories:
- **Convergent:** Toward expected profiles → **Transient immaturity**.
- **Persistent Atypical:** Despite adjustments → **Enduring constraints** (Nigg, 2017).

3. The Dual Role of Early Intervention:

- Early intervention serves a **therapeutic role** AND a **diagnostic function**.
- It enables targeted adjustments to distinguish **transient** difficulties from **persistent** constraints

3.Developmental and Neurobiological Foundations

1. Brain Maturation:

- **Non-linear and asynchronous** across regions.
- **Hierarchical trajectory** (caudo-rostral progression).
- Sensory/motor regions mature early; prefrontal regions develop into adulthood.
- High **intra- and inter-individual variability**.

2. Clinical Implication:

- Absence of a skill may reflect **transient maturation**, not a stable disorder.
- **High plasticity** in childhood allows adaptation but increases **sensitivity to context**.
- Plasticity unfolds within **constrained trajectories** (sensitive periods).

3. Evolving Environmental Demands:

- Contexts become increasingly complex.
- Difficulty visibility depends on the **mismatch** between individual resources and environmental demands.
- Some difficulties emerge only when the balance is no longer sufficient.
- Symptoms must be interpreted **relative to developmental expectations**.

4. The Structuring Role of Time:

- Time is not a background variable but a **structuring dimension** in neurodevelopment

Age-Appropriate Assessment & The Risk of Misinterpretation

1. Age-Appropriate Assessment:

- Developmental changes influence the interpretation of neurological and behavioral findings (Prechtl, 2001).
- **Age-specific constellations** of features may facilitate earlier detection (Marschik et al., 2017).

2. The Relative Age Effect :

- The youngest children in a school cohort are **more likely to receive an ADHD diagnosis** (Caye et al., 2020).
- This may reflect **relative developmental immaturity**, not qualitative differences in functioning.
- **Clinical Risk:** Misinterpreting typical developmental variation as pathological when the **temporal dimension** is insufficiently considered.

3. Time as a Clinically Informative Variable:

- Time is **not merely a background dimension**; it is a **clinically informative variable**.
- Its interpretation depends on **developmental timing** and **age-appropriate expectations**.

4. The Key Question:

- If developmental heterogeneity is intrinsic to neurodevelopment, how can this **temporal dimension** be incorporated into diagnostic reasoning and clinical decision-making?

۴.Time and Developmental Trajectories in Diagnostic Reasoning

1. Longitudinal Follow-Up:

Provides a framework for analyzing **developmental trajectories** (Marschik et al., 2017).

2. Current Diagnostic Reasoning vs. Prospective Observation:

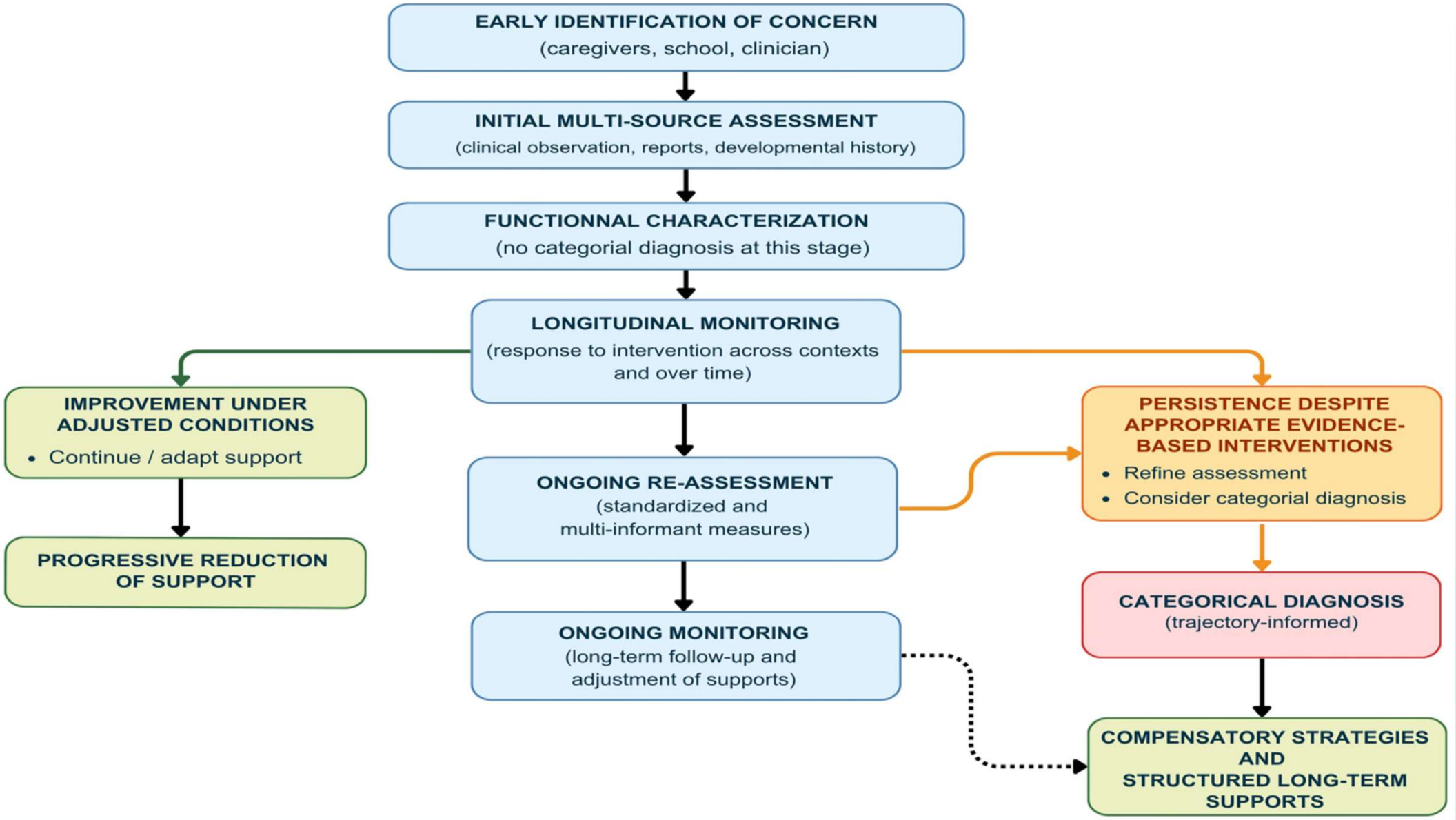
Current: Relies primarily on **current observations** and **retrospectively reconstructed accounts**.

Key Insight: Understanding how difficulties **emerge, evolve, or persist** as environmental conditions change is essential.

3. Interventions as Diagnostic Tools:

Interventions are **not only therapeutic**; they reveal **developmental responses**

4. Illustrative Example: Developmental Language Trajectories:~10-20% of 2-year-olds show **late language emergence**.Longitudinal studies: **~75-85%** attain language abilities within the average range.Only a **minority** present persistent difficulties at school age


EARLY IDENTIFICATION OF CONCERN
(caregivers, school, clinician)
INITIAL MULTI-SOURCE ASSESSMENT
(clinical observation, reports, developmental history)
FUNCTIONNAL CHARACTERIZATION
(no categorial diagnosis at this stage)
LONGITUDINAL MONITORING
(response to intervention across contexts and over time)
IMPROVEMENT UNDER ADJUSTED CONDITIONS
• Continue / adapt support
PROGRESSIVE REDUCTION OF SUPPORT
ONGOING RE-ASSESSMENT
(standardized and multi-informant measures)
ONGOING MONITORING
(long-term follow-up and adjustment of supports)
PERSISTENCE DESPITE APPROPRIATE EVIDENCE-BASED INTERVENTIONS
• Refine assessment
• Consider categorial diagnosis
CATEGORICAL DIAGNOSIS
(trajectory-informed)
COMPENSATORY STRATEGIES AND STRUCTURED LONG-TERM SUPPORTS

5. Executive Functions as Illustrative Developmental Markers

1. Why Executive Functions (EF)?

- A **transdiagnostic developmental construct** (Nigg, 2017; Roy and Fournet, 2021).
- Involved across neurodevelopmental, psychiatric, neurological, and genetic conditions.
- Associated with **heterogeneous manifestations** affecting social, emotional, and behavioral functioning.
- Documented in ASD, language disorder, prematurity, brain injury, and genetic conditions.
- Involved in **compensatory strategies and adaptive functioning**.

2. Developmental Trajectory of EF:

- **Prolonged trajectory** (early childhood to adulthood).

- Early forms from preschool; specialization with maturation of **fronto-subcortical networks**.
- Difficulties become visible when **environmental demands exceed regulatory capacities**.

3. EF as a Developmental Window:

- Rather than disorder-specific markers, a **developmental window** onto regulatory processes.
- Insight into the **dynamic balance** between brain maturation, resources, compensatory strategies, and environmental demands.
- Expression varies across contexts → **point-in-time interpretations uncertain**.
- **Repeated assessments** are therefore needed.

5.Executive Functions as a Developmental Lens

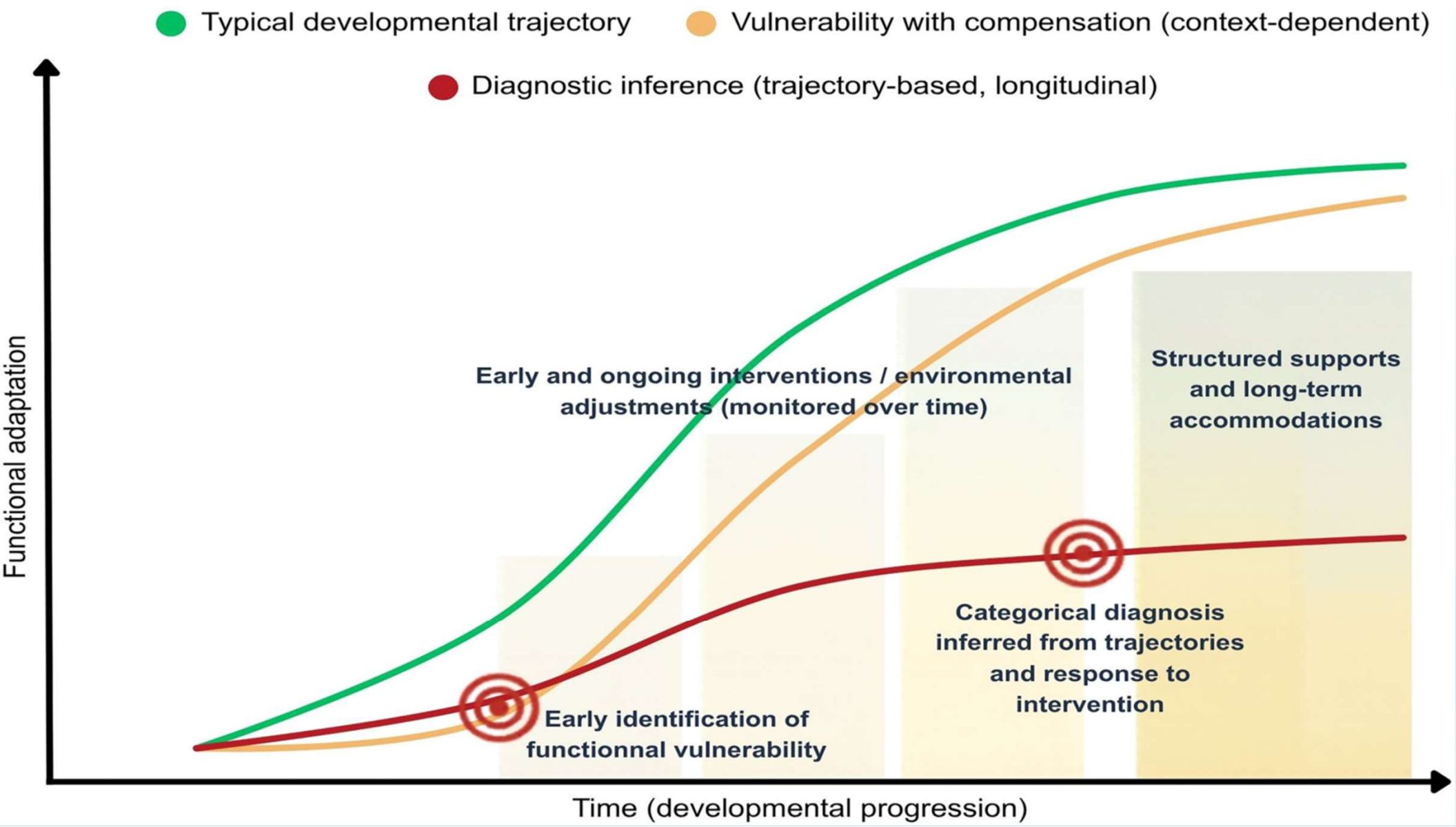
1. Interpreting Response to Support:

- **Improvement** following structured support → suggests a **contextual imbalance**.
- **Persistence** despite appropriate interventions → supports the hypothesis of an **underlying neurodevelopmental constraint**.

2. Tracking EF Over Time:

- Tracking EF over time and across contexts helps identify **persistent deviations**.
- This suggests **more durable neurodevelopmental constraints** (Figure 2).

3. EF as a Developmental Lens


Typical developmental trajectory
Vulnerability with compensation (context-dependent)
Diagnostic inference (trajectory-based, longitudinal)
Functional adaptation
Early and ongoing interventions / environmental adjustments (monitored over time)
Structured supports and long-term accommodations
Categorical diagnosis inferred from trajectories and response to intervention
Early identification of functionnal vulnerability
Time (developmental progression)

.۶ Clinical Implications for longitudinal diagnostic reasoning

1. The Limitation of Cross-Sectional Assessment:

- A single assessment (consultation, test, or report) cannot fully capture the developmental process.
- Diagnostic value is **limited** if not interpreted in relation to **temporal evolution** and **responsiveness to environmental adjustments**.

2. The Solution: Structured Longitudinal Monitoring:

- Complement evaluations with **repeated assessments over time**.
- **This does NOT mean delaying diagnosis.**
- It means **starting early intervention now**, while **refining the diagnosis over time**.

3. The Role of Clinical Judgment:

- Clinical judgment remains central but must rely on **longitudinal data**.
- **Tools:** Ecological observations, multi-informant questionnaires, adaptive measures, repeated standardized assessments.

4. Key Analogy:

- Developmental assessment is like **growth monitoring** — you don't measure a child's height just once.

Limitations and Future Directions

- Does **NOT** propose a new diagnostic framework.
- Aims to make **explicit** existing practices (trajectories, longitudinal reasoning)
- **Future Directions:**
- No consensus on incorporating supported trajectories into diagnostic reasoning.
- Need **reliable developmental indicators** (stability, malleability, responsiveness).
- Strengthen clinician's role in **individualized diagnostic formulations** (not just single-time-point symptoms).


از توجه شما متشکرم