Director's Report on Developmental Progress Assessment
Date of Visit: 07-05-2025
Child's Name: Faisal Amro Abu Altahir
Date of Birth: 2017-01-18

Overview:
Faisal was assessed at our center and presents with **information processing delays**, **intellectual developmental delays**, and **acquired features of autism**. A detailed genetic analysis (Whole Exome Sequencing via Neurazon Canada) revealed multiple **variants of unknown significance (Class 3)**, with a special emphasis on the **SCN2A gene**, which is of high clinical relevance in this case.

Among the variants identified, the **SCN2A c.1837G>C (p.Val613Leu)** heterozygous mutation stands out due to its well-documented involvement in **SCN2A-related neurodevelopmental disorders**, including autism, epilepsy, and cognitive impairment. SCN2A encodes a voltage-gated sodium channel subunit critical for neuronal excitability and signal transmission. Dysfunction in this channel often results in altered cortical activity and impaired synaptic development, both of which are consistent with Faisal's current developmental profile.

Other variants found include:

- **ADGRL1**: Implicated in developmental and neuropsychiatric conditions.
- **SLC33A1**: Linked to spastic paraplegia.
- **MYO7A**: Recently recognized as a candidate gene in autism due to its association with actin-based transport in neuronal structures.

While none of the variants are classified as definitively pathogenic, their collective presence may synergistically contribute to Faisal's phenotype. Clinical correlation and regular reassessment are essential.

Recommended Plan:

1) Development Protocol for Information Processing

- Initiate a **non-pharmacological neurodevelopmental protocol** focused on enhancing information processing and cognitive function.
- Duration: **4 months**
- Reassessment: Reevaluate the child in person to measure progress and establish a data-driven baseline for future intervention.
- Continue with structured therapy sessions and cognitive stimulation activities.

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2) Genetic and Biomedical Considerations

- The **SCN2A variant** should be monitored for potential seizure onset or atypical neurological events. We recommend **neurology consultation** and **EEG screening** if clinically warranted.
- Family genetic counseling is advised due to the autosomal dominant inheritance pattern of the identified variants.
- Maintain ongoing evaluation of developmental, behavioral, and cognitive functions every 3–4 months.

Conclusion:
Faisal's developmental profile and genetic findings support the diagnosis of acquired features of autism and intellectual delays influenced by underlying genetic susceptibilities, particularly in the **SCN2A pathway**. While this is not a final medical diagnosis, the assessment provides a critical foundation for tailored intervention and supports the need for continued therapeutic and clinical oversight.

Signature:
EICADD Center Director