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Medical report

Patient: Hamid Swaleh Omar
Age: 8 years 9 months
Date of First Evaluation: At 7.5 years
Prepared by: Dr Farida Essajee

Background History

- **Pregnancy & Birth:** Normal pregnancy. Failed induction, prolonged labor, delayed cry at birth. Birth weight: 2.9 kg. Initial feeding difficulties with poor swallowing, reflux, and poor weight gain.
- **Developmental History:**
  - Early delays noted. Sat late with stiffness.
  - Achieved gross motor milestone of walking but with clumsiness.
  - Fine motor coordination remained poor.
  - Speech development staggered.
  - Drooling noted.
- **Regression:** Previously able to eat independently and support himself, now with progressive fatigue and slow decline in function.
- **Schooling:** Enrolled in mainstream school. Concentration noted to be poor but overall performance satisfactory. Now homeschooling as child cant cope with walking long distances.
- **Past Medical History:** No history of seizures. Toilet trained. Weak immune system with recurrent infections and poor weight gain. No cardiac involvement reported.
- **Family History:** Mother with cervical spondylosis-related hand weakness.

Clinical Examination (Initial Review)

- **Anthropometrics:** Weight 15.7 kg; Length 106.5 cm; Head circumference 49 cm.
- **Neurological:**
  - Oculomotor apraxia.
  - Normal upper limb tone and reflexes.
  - Lower limbs: hypotonia, ankle reflexes absent.
  - Romberg's sign positive.
  - Ataxia absent at the time.
  - Cognitive skills age-appropriate.

Investigations Ordered: MRI brain, FBC, TSH, Alpha-fetoprotein (AFP), CK, Lactate.

Follow-Up Review & Results

- **Investigations:** Revealed **cerebellar atrophy** and **markedly elevated alpha-fetoprotein**, consistent with **Ataxia Telangiectasia (A-T)**.
- **Anthropometrics:** Weight 17 kg; Height 111.5 cm.
- **Vitals:** HR 125/min; SpO_2 98%.

Neurological Examination

- Oculomotor apraxia with ocular telangiectasia.
- Tone and reflexes: Upper limbs normal.
- Ataxia present: past pointing, intention tremor, ataxic gait.
- Chorea, more pronounced in the upper limbs.
- Romberg's sign positive.
- Lower limb strength preserved; knee reflexes present, ankle reflexes absent.
- Vibration sense absent.
- Minimal drooling.

Current Assessment

Diagnosis: *Ataxia Telangiectasia* (confirmed on clinical grounds with raised AFP and cerebellar atrophy).
This is a rare, progressive neurodegenerative condition characterized by:

- Cerebellar ataxia and movement disorders (tremor, chorea).
- Oculomotor apraxia.

- Immunodeficiency leading to recurrent infections.
- Poor weight gain and growth failure.

Management Plan

1. **Medications & Supplements:**
   - **Coenzyme Q10** 100 mg once daily.
   - **Vitamin E supplementation.**
   - **Amantadine** 25 mg once daily for tremor and movement disorder.
2. **Therapy & Rehabilitation:**
   - Referral to pediatric occupational therapy for ataxia and functional support.
   - Chest physiotherapy as needed to support respiratory function.
3. **Further Recommendations:**
   - Enrolment in **clinical trials for gene therapy** where accessible.
   - Regular neurological and immunological follow-up.
   - Continued supportive school environment with adaptations for motor fatigue.

Sincerely,

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