

Collaborating care in rare genetic disorders- a case study

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Who are we?

- **RHS established 1999**
- **Statewide service- Liverpool, Armidale, Coffs Harbour, Newcastle, Wollongong & Wagga Wagga**
- **“Protect & promote health of refugees & people of refugee like backgrounds”**
- **DST established 2017**

Nursing approach – The how & why

- **Culturally responsive**
- **“Disability”- Introducing concepts**
- **Health literacy**
- **Trauma-informed model of care**
- **Impacts of experiences-
empowerment to make decisions**

Case Study- Maple Syrup Urine Disease (MSUD)

- **6-year-old boy born in Ankara, Turkiye**
- **Afghan family, Dari speaking**
- **Rare genetic disorder of enzymes which metabolize branch-chain amino acids (BCAAs)**
- **Prevalence- 1:185,000**
- **Usually seen in 12-48 hours of life**
- **Characterized by sweet “maple syrup like” smell in urine**

Clinical presentation

- **Metabolic abnormalities, ketonuria**
- **Neurological dysfunction/oedema**
- **Poor feeding/intake**
- **Vomiting, gastro**
- **Seizures & abnormal muscle movements**
- **Irreversible neurocognitive deficit, coma, loss of life**

Diagnosis & beyond

- **Metabolic correction**
- **Hydration, formula/supplements**
- **Restriction of BCAAs**
- **Neurovascular support, prevention of encephalitis/seizures, D&V**
- **ICU management**

Long term:

- **Strict dietary intake (protein)/supplemental feeds**
- **Regular metabolic screening/specialised clinicians**
- **Education to avoid crisis/plan**
- **Prevention of long-term effects**

The picture

- **Diagnosed aged 4 days old**
- **Classic symptoms, decompensated**
- **Long ICU admission**
- **Neurological injury**
- **Seizures/epilepsy**
- **Global developmental delay**

Planning for arrival

Early coordination:

- Refugee Nurse assessment
- Health Assessment for Refugee Kids/Westmead Kids
- Settlement Services International (SSI)
- DST
- NDIS Early Childhood
- STARTTS (Service for the Treatment and Rehabilitation of Torture and Trauma Survivors)

Looking ahead

- **Discharge planning**
- **Brain Injury Clinic, Orthopedics, specialist dental, rehabilitation (allied health)**
- **NDIS process/ Early Childhood**
- **Family settlement complexities**

Where are we now?

9 months since arrival

- Long term accommodation
- Starting school
- Dad enrolled in English language course
- NDIS plan- therapies & equipment

Conclusions

- **Partnership is vital- nurture connections**
- **Nurses are central to the process**
- **Recognition of value**
- **Health equity- navigation**
- **Advocacy is key**

Links/information

- NSW Refugee Health Service - www.swslhd.health.nsw.gov.au/refugee
- STARTTS (Service for the Treatment and Rehabilitation of Torture and Trauma Survivors) - www.startts.org.au
- United Nations High Commission for Refugees - www.unhcr.org/au
- NSW Refugee Health Plan 2022/2027
- www.health.nsw.gov.au/multicultural/Pages/refugee-health-plan
- National Organization for Rare Diseases (NORD)
<https://rarediseases.org/rare-diseases/maple-syrup-urine-disease>

References

- **Maple Syrup Urine Disease-** Hassan, SA & Gupta, National Library of Medicine, National Institute of Health, 2022
- **Trauma-informed Care: A sociocultural perspective, Trauma-Informed Care in Behavioral Health- Services,** Rockville, 2014

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- **NSW Refugee Health Service** - www.swslhd.health.nsw.gov.au/refugee
- **STARTTS (Service for the Treatment and Rehabilitation of Torture and Trauma Survivors)** - www.startts.org.au
- **United Nations High Commission for Refugees** - www.unhcr.org/au
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