**Supplementary table 1: Literature review of the previously reported patients with GS2 in the MENA region.**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **No. of patients** | **Consanguinity** | **Cutaneous findings** | **CNS findings** | **HLH** | **RAB27A pathogenic variant** | **Origin** | **Reference** |
| 12 | 12/12 | 12/12 | 7/12 | 7/12 | N/A | Arabian  Peninsula | (Harfi et al., 1992) |
| 1 | + | + | + | - | p.S18WfsX15 | Arabic | (Aksu et al., 2003) |
| 2 | + | + | + | + | N/A | Iran | (Ashrafi et al., 2006)  (2t al |
| + | + | N/A | N/A | N/A |
| 1 | + | + | + | - | p.K134E | Jordan | (Masri et al., 2008) |
| 9 | 9/9 | N/A | 4/9 | 6/9 | * p.Gln172fsX1 (5/9) * p.Ile114X (1/9) * p.Ile44Thr (1/9) * p.Arg50fsX34+ and p.Arg50Ther (1/9) * P.Gln172fsX1 (1/9) | Iran | (Mamishi et al., 2008) |
| 1 | + | + | - | + | p.K22R | Iran | (Ohbayashi et al., 2010) |
| 1 | + | + | - | + | p. S115G | Iran | (Shamsian et al., 2010) |
| 2 | - | + | + | - | p.F46I | India | (Mishra et al., 2014) |
| - | + | - | + | p.F46I |
| 4 | + | + | + | + | p.Arg50GlnfsX35 | Turkey | (Patiroglu et al., 2016) |
| + | + | - | - | p.Arg50GlnfsX35 |
| + | + | - | + | p.Gln172AsnfsX2 |
| + | + | - | + | p.Gln172AsnfsX2 |
| 1 | N/A | N/A | N/A | + | p.R82C | China | (Jin et al., 2018) |
| 35 | 33/35 | N/A | 16/35 | 28/35 | 20/35   * RAB27A tandem duplication (3/20) * p.R50fsX33 (6/20) * p.R82C (3/20) * p.R200X (3/20) * p.Q172X (2/20) * p.W73X (1/20) * p.K144Q (1/20) * p.A92fsX7 (1/20) | KSA | (Al-Mofareh et al., 2020) |