**Introduction**

The mucopolysaccharidoses (MPSs) are a group of rare, life-limiting lysosomal storage diseases. Prompt diagnosis is essential for early access to appropriate care and where disease modifying treatment is available, to prevent irreversible tissue and organ damage.1,2

In the UK, MPSs are not included in newborn screening panels and due to the rarity of these conditions, diagnosis can take a number of years from the point where parents first raise their concerns to a healthcare provider.2

**Aim**

We conducted a study to determine the age at diagnosis for MPS in the UK.

**Methods**

The MPS Society UK has systematically collected data on its members since 1987 and holds a database containing records on over 900 MPS patients.

- This database was searched in August 2020 for all records of patients with an MPS disease
- Patients with missing data were identified (no date of birth or no date of diagnosis)
- MPS Society archives were searched and patients or family contacted to fill data gaps

**Patient numbers**

Date of birth and date of diagnosis was available for 760 MPS patients in total (Table 1).

<table>
<thead>
<tr>
<th>Type of MPS</th>
<th>Number of patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>MPS I</td>
<td>201</td>
</tr>
<tr>
<td>MPS II</td>
<td>155</td>
</tr>
<tr>
<td>MPS III</td>
<td>272</td>
</tr>
<tr>
<td>MPS IV</td>
<td>90</td>
</tr>
<tr>
<td>MPS VI</td>
<td>41</td>
</tr>
<tr>
<td>MPS VII</td>
<td>1</td>
</tr>
</tbody>
</table>

**Table 1. Number of patients by type of MPS**

**Results**

**Age at diagnosis**

The earliest date of diagnosis recorded was in 1966. The age at diagnosis ranged from pre-natal to 52 years. Variability was seen between the different types of MPS (Figure 1):

- Overall, those with MPS I were diagnosed earlier than patients with other types of MPS
- Median age at diagnosis for MPS II and MPS IVA was similar and at an earlier age than MPS VI
- MPS III patients were generally older at diagnosis than those with other types of MPS
- The MPS VII patient was diagnosed early due to the presence of hydrops fetalis

**Diagnosis in the last 30 years**

The MPS Society held records for 623 patients who had been diagnosed since 1990. Over this period there was a slight trend towards diagnosis at an earlier age (Figure 4).

**Conclusions**

There has been very little change in the age at diagnosis of MPS over the last 30 years. This illustrates the difficulty healthcare providers have in recognising the early signs of these rare diseases. With the continued development of disease modifying treatments that have the potential to address the devastating effects of MPS if treatment is started early enough, there is a continued need to identify these patients at an earlier age.