JIA-LIKE IN A BOY WITH ATAXIA-TELANGIECTASIA
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Introduction
Ataxia-Telangiectasia (A-T) is an autosomal recessive disorder, caused by mutations in the ATM-gene leading to misfunction of ATM, a protein kinase that is involved in double stand breaks-damage-sensing and cell-cycle-arrest, crucial for DNA-repair. Cells with higher turn-over rates such as neurological and immune cells, are more susceptible to DNA damage, rendering A-T patients to a degenerative, progressive disease consisting in neurological impairment, increased susceptibility to malignancy and predisposition to infections. Despite being linked with many primary immunodeficiencies (PID), juvenile idiopathic arthritis (JIA) has rarely been associated with DNA-repair disorders, such as A-T.
We report a case of JIA-like in a male patient with A-T.

Case Report

Birth

Healthy during the first year of life

1 year

Pain and swelling of 4th metatarsophalangeal (MTP) joint of left foot

2 years

Stars walking – unstable gait

Two disease associated mutations of ATM gene

3 years

After 6 months symptoms persisted

4 years

Primary immunodeficiencies

Rheumatology

C.3894 dup-exon 26
C.5291T>C-exon 35

Ataxia-Telangiectasia

Table 1 – Immunologic evaluation

<table>
<thead>
<tr>
<th></th>
<th>At diagnosis</th>
<th>2.5 YO</th>
<th>5 YO</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>IgA (g/L)</td>
<td>&lt;0.23</td>
<td>&lt;0.23</td>
<td>0.33-3.60</td>
<td></td>
</tr>
<tr>
<td>IgM</td>
<td>1.56</td>
<td>1.74</td>
<td>0.55-2.10</td>
<td></td>
</tr>
<tr>
<td>IgG</td>
<td>11.30</td>
<td>10.2</td>
<td>5.93-17.30</td>
<td></td>
</tr>
<tr>
<td>IgE</td>
<td>&lt;2.0</td>
<td>&lt;2.0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lymphocytes (l/mm3)</td>
<td>1870</td>
<td>1740</td>
<td>3000-9500</td>
<td></td>
</tr>
<tr>
<td>CD3+ (cells/µl)</td>
<td>545</td>
<td>485</td>
<td>1400-3700</td>
<td></td>
</tr>
<tr>
<td>CD4+ (%CD3CD45RA)</td>
<td>300 (7%)</td>
<td>274 (9%)</td>
<td>700-2200</td>
<td></td>
</tr>
<tr>
<td>CD8+ (%CD3CD45RACD62)</td>
<td>131 (28%)</td>
<td>133 (40%)</td>
<td>490-1300</td>
<td></td>
</tr>
<tr>
<td>CD19+</td>
<td>183</td>
<td>154</td>
<td>390-1400</td>
<td></td>
</tr>
<tr>
<td>NK-cells (cells/µl)</td>
<td>348</td>
<td>386</td>
<td>130-720</td>
<td></td>
</tr>
<tr>
<td>CD3+TCRγδ (%)</td>
<td>144 (29%)</td>
<td>170 (30%)</td>
<td>1-10%</td>
<td></td>
</tr>
</tbody>
</table>

Vbeta repertoire: oligoclonal, restricted
Abnormal proliferative lymphocyte response to mitogens and recall Antigens

Arthritic of:
• Both knees
• Left ankle
• 4th MTP of the left foot
Tenosynovitis of:
• 2nd finger of the right hand

Table 2 – Laboratory investigations

| Erythrocyte sedimentation rate | 25 mm/h |
| C-reactive protein            | 1.45 mg/L |
| Antinuclear antibodies        | positive (1/80) |
| Anti-cyclic citrullinated protein | negative |
| Rheumatoid factor             | negative |

Remain active:
Arthritis of 4th MTP of left foot
Tenosynovitis of 2nd finger of right hand

Hydroxichloroquine
(5 mg/kg/day)

JIA-like

Ibuprofen (30 mg/kg/day)
Chemical synovectomy with triamcinolone hexacetonide (TH)
(knees and ankle)

Terapeutic ??

Methotrexate?

Conclusion
This case seems to be the first known pediatric patient with A-T who developed chronic, JIA-like oligoarthritis. The management of these patients is particularly difficult because they are extremely susceptible to DNA damage and show an increased susceptibility to viral infections (namely herpetic). These features are very important when considering the best therapeutic options for JIA-like arthritis.

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References: