Shwachman-Diamond Syndrome: First Successful Hematopoietic Stem Cell Transplant in Nebraska

Grace Murray et al.

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The use of two well-matched UCB units has treated disorder with medication, Stanford-Binet Patient demographics and clinical characteristics.

**Table 1.**

<table>
<thead>
<tr>
<th>By Case (n=47)</th>
<th>By Patient (n=14)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>N (%)</td>
</tr>
<tr>
<td>Male</td>
<td>17 (36.1)</td>
</tr>
<tr>
<td>Female</td>
<td>30 (63.8)</td>
</tr>
<tr>
<td>Age</td>
<td></td>
</tr>
<tr>
<td>0-5</td>
<td>19 (40.4)</td>
</tr>
<tr>
<td>6-10</td>
<td>6 (12.7)</td>
</tr>
<tr>
<td>11-15</td>
<td>11 (23.4)</td>
</tr>
<tr>
<td>16-20</td>
<td>8 (17)</td>
</tr>
<tr>
<td>21+</td>
<td>3 (6.4)</td>
</tr>
<tr>
<td>Average</td>
<td>Mean 10 y</td>
</tr>
</tbody>
</table>

Clinical Characteristics

<p>| | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Seizure history</td>
<td>5 (35.7)</td>
</tr>
<tr>
<td>Contractures</td>
<td>14 (100)</td>
</tr>
<tr>
<td>Photophobia</td>
<td>6 (42.8)</td>
</tr>
<tr>
<td>Able to ambulate</td>
<td>10 (71.4)</td>
</tr>
<tr>
<td>IQ</td>
<td></td>
</tr>
<tr>
<td>≤50</td>
<td>7 (50)</td>
</tr>
<tr>
<td>&gt;50</td>
<td>6 (42.8)</td>
</tr>
</tbody>
</table>

Mean=55 Median=50.5

*Names in bold type indicate presenting author.

Double Umbilical Cord Blood Transplantation in a Pediatric Patient: A First for Nebraska
Grace Murray1, Sachit Patel1
1University of Nebraska Medical Center, Department of Pediatrics

Mentor: Sachit Patel
Program: Pediatrics
Type: Case Report

Background: Unrelated umbilical cord blood (UCB) transplantation has been used as a hematopoietic stem cell source for 30 years. Compared to adult bone marrow and peripheral blood stem cells, UCB has more rapid availability, absence of donor attrition, and reduced risk of GVHD despite HLA disparity. Unfortunately for larger patients, a single UCB unit has an insufficient amount of total nucleated cells to support engraftment. The use of two well-matched UCB units has been shown to overcome this barrier.

Methods: Chemotherapy, Graft, Cord Blood Transplant. Consent was obtained to utilize this case for educational purposes.

Results: A 12 year-old presented with bleeding and weight loss and was found to have anemia, thrombocytopenia and leukocytosis. Peripheral smear demonstrated auer rods. He was diagnosed with acute myeloid leukemia (AML-M4). He successfully completed therapy with protocol AAML 1031 but relapsed seven months later. He achieved a second complete remission on day +27. He is now 16 years-old with full donor chimerism and complete immune reactivation. Engraftment was achieved on day +42. Complications included grade III aGVHD involving the skin and GI system, and CMV reactivation. General anesthesia was well tolerated.

Conclusion: Patients with SLS typically require general anesthetics when undergoing diagnostic studies and procedures for symptom management. Our case series suggests that general anesthesia is well tolerated in this population. Important aspects of pre-anesthetic evaluation include history of seizure severity and control, neurocognitive assessment and skin examination. Intraoperative considerations include photophobia, difficulty securing IVs and monitors, relative heat intolerance, hypohydrosis, and challenges with positioning, padding, and line placement due to contractures. The ichthyosis of SLS spares the midface eliminating difficulty in securing airway devices. https://doi.org/10.32873/unmc.dc.gmerj.2.1.068

Shwachman-Diamond Syndrome: First Successful Hematopoietic Stem Cell Transplant in Nebraska
Grace Murray1, Sachit Patel1
1University of Nebraska Medical Center, Department of Pediatrics

Mentor: Sachit Patel
Program: Pediatrics
Type: Case Report

Background: Shwachman-Diamond syndrome (SDS) is an autosomal recessive condition characterized by bone marrow dysfunction, pancreatic insufficiency, and skeletal abnormalities. Ninety percent of patients with SDS have a mutation in the SBDS gene on chromosome 7, while the other ten percent are diagnosed clinically.

Methods: Chemotherapy, Graft, Cord Blood Transplant. Consent was obtained to utilize this case for educational purposes.

Results: A 12 year-old presented with bleeding and weight loss and was found to have anemia, thrombocytopenia and leukocytosis. Peripheral smear demonstrated auer rods. He was diagnosed with acute myeloid leukemia (AML-M4). He successfully completed therapy with protocol AAML 1031 but relapsed seven months later. He achieved a second complete remission with protocol AAML 0523. Given his high risk disease and poor prognosis with chemotherapy alone, the patient was offered transplant. No suitable sibling or unrelated donor donors were found. He underwent mismatched double umbilical cord transplant.

Conclusion: This was the first pediatric patient in Nebraska to have a successful double UCB transplant. Double cord transplant is an acceptable alternative when there is no sibling or unrelated donor match and when a single cord unit total nucleated dose is insufficient. https://doi.org/10.32873/unmc.dc.gmerj.2.1.069
Acute Onset Parkinson's Disease Secondary to West Nile Virus Encephalitis

Matthew Purbaugh1, Fuad-al Ali1, T. Scott Diesing1, Amy Hellman1
1University of Nebraska Medical Center, Department of Neurological Sciences

Methods: Bone Marrow Biopsy, Transplant. Consent was obtained to use this case for educational purposes.

Results: A 2 month-old male presented with pallor, diarrhea and anemia. He subsequently developed thrombocytopenia and neutropenia. A bone marrow biopsy was performed and demonstrated hypocellular bone marrow. Parents reported a history of oily stools and lab findings were suggestive of pancreatic insufficiency. The patient had an inconclusive genetic work-up but was clinically diagnosed with SDS. The patient was weekly transfusions at 4 years-old. Due to his transfusion dependence, he was determined to be a candidate for HSCT. The patient received a novel reduced intensity conditioning followed by an allogeneic mismatched unrelated bone marrow transplant. Complications following transplant included mucositis, moderate veno-occlusive disease, and disseminated adenovirus. He is now 6 years-old, fully engrafted, off immunosuppression, and transfusion-independent.

Conclusion: Our patient is the first in Nebraska with SDS to receive a successful HSCT. Early recognition and diagnosis of the disease can decrease the incidence of MDS/AML and transfusion-related complications. In SDS patients who progress to transplant, reduced intensity conditioning can decrease transplant-related morbidity and mortality and improve long-term quality of life.

https://doi.org/10.32873/unmc.dc.gmerj.2.1.070

References

Acute onset Parkinsonism

Neuroinvasive West Nile Virus: A Case Series in Nebraska

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1University of Nebraska Medical Center, Department of Neurological Sciences

Methods: We reviewed charts of four patients admitted to neurology service who had positive serology and/or polymerase chain reaction (PCR) testing for WNV.

Results: Case 1: A 39-year-old female with diffuse large B-cell lymphoma presented with fever, encephalopathy, and flaccid paraplegia. MRI showed T2 hyperintensities involving the bilateral thalami, temporal lobes, midbrain, pons, and spinal cord which were suspicious for CNS lymphoma. WNV was confirmed by PCR as her prior treatment with RCHOP made her serum and CSF serology falsely negative. Case 2: A 49-year-old female presented with left sided paresis and left arm weakness. WNV was confirmed by CSF lymphocytic pleocytosis and positive WNV IgM. Case 3: A 79-year-old male presented with acute onset parkinsonism with positive CSF WNV IgM. Case 4: A 31-year-old male who presented with uveo-meningitis syndrome with uveitis of the right eye, fever, headache, nuchal rigidity and CSF pleocytosis. An autoimmune cause was suspected until CSF showed WNV IgM. All four patients showed varying amounts of neurological recovery with supportive care.