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CHALLENGES OF SMITH-LEMLI-OPITZ SYNDROME IN THE NEONATAL INTENSIVE CARE UNIT: A CASE REPORT

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Background: Smith-Lemli-Opitz syndrome (SLOS) is a rare autosomal recessive genetic disorder caused by disrupted cholesterol biosynthesis, specifically a deficiency of the enzyme 7-dehydrocholesterol reductase (DHCR7), which converts 7-dehydrocholesterol (7DHC) to cholesterol. Multiple congenital anomalies, psychomotor developmental delay, and behavioral disturbances characterize SLOS.

Case presentation: A newborn delivered at 39 weeks of gestation presented with multiple dysmorphic features and congenital anomalies, including microcephaly, low-set ears, micrognathia, epicanthal folds, hypertelorism, syndactyly, hypospadias, unilateral cryptorchidism, a high-arched palate, a narrow forehead, ptosis, downturned corners of the mouth, thin lips, hypotonia, hyporeflexia, and constipation. On the 12th day of life, the infant developed projectile vomiting. Abdominal ultrasonography revealed pyloric stenosis, and laboratory investigations confirmed bacterial sepsis. Antibacterial therapy was initiated, and the patient underwent surgical treatment with pyloromyotomy. During follow-up, the vomiting resolved, along with the clinical and laboratory signs of infection. However, feeding intolerance and poor weight gain persisted. Genetic testing was subsequently performed using whole-exome sequencing (WES), which identified two pathogenic variants in the DHCR7 gene: c.278C>T, p.(Thr93Met), and c.964-1G>C, p.(?). These findings confirmed the diagnosis of Smith-Lemli-Opitz syndrome (SLOS). Smith-Lemli-Opitz syndrome is characterized by impaired cholesterol biosynthesis, resulting in low serum total cholesterol levels and elevated levels of 7-dehydrocholesterol (7-DHC). Following the diagnosis, in addition to a high-calorie diet, the patient was administered daily cholesterol supplementation at a dose of 4 g/day. Subsequently, the infant demonstrated weight gain and a relative improvement in feeding tolerance.

Conclusion: This case highlights the importance of considering Smith-Lemli-Opitz syndrome (SLOS) in the differential diagnosis of male newborns presenting with severe feeding difficulties accompanied by multiple dysmorphic features and congenital anomalies. Therefore, in infants with poor weight gain, assessment of total serum cholesterol and the lipid profile should be

considered, along with molecular genetic testing, which can help establish the underlying genetic cause of the clinical presentation and confirm the diagnosis.

Keywords: Smith-Lemli-Opitz syndrome; feeding intolerance; poor sucking; poor weight gain; total cholesterol; lipid profile.