GRISCELLI SYNDROME, TYPE 2

Alternative Name
GS2
Griscelli Syndrome with Hemophagocytic Syndrome
Partial Albinism and Immunodeficiency Syndrome
Paid Syndrome

Record Category
Disease phenotype

WHO-ICD
Endocrine, nutritional and metabolic disease > Metabolic disorders

Incidence per 100,000 Live Births
NA

OMIM Number
607624

Mode of Inheritance
Autosomal recessive

Gene Map Locus
15q21.3

Description
Griscelli syndrome is an inherited condition characterized by hypopigmented skin, silver-gray hair, neurological and immune system abnormalities. There are three types with different signs and genetic causes. Patients with type 2 are prone to recurrent infections and they develop hemophagocytic lymphohistiocytosis (HLH). The prevalence is <1 / 1 000 000. The onset of the signs of the disease is in infancy and death happens in childhood if untreated. Diagnosis can be made by clinical presentation, hair shaft study, imunological laboratory finding and genetic tests. Treatment includes bone marrow transplantation.

Molecular Genetics
RAB27A is located on the long arm of chromosome 15. Mutations in RAB27A causes abnormal pigmentation of hair and skin, resulting in Griscelli syndrome type 2. This gene contains 5 coding exons. Several mutations have been described.

Epidemiology in the Arab World
Saudi Arabia

Al-Ahmari et al., (2010) studied 11 patients with GS who received allogeneic hematopoietic stem cell transplants (aHSCT) between 1993 and 2007. The median age at transplantation was 8.2 months. Seven patients treated with chemotherapy before transplantation because they reached accelerated phase. Two sources of grafts were available; matched-related marrows in 8 patients and mismatched unrelated cords in the remaining three. Fifteen days was the median time for engraftment in all subjects. Ten patients were alive at a median age of 4.8 years after transplantation. One patient died of septic shock 6 months after transplantation. The authors concluded that early aHSCT is feasible for treating patients with Griscelli syndrome and low level of donor cell engraftment is sufficient to prevent the recurrence of the disease.

References

A Al-Ahmari, A Al-Ghonaium, M Al-Mansoori, A Hawwari, A Eldali, M Ayas, H Al-Mousa, A Al-Jefri, B Al-Saud, A Al-Seraihy, S Al-Muhsen, M Al-Mahr, H Al-Dhekri and H El-Solh. Hematopoietic SCT in children with Griscelli syndrome: a single-center experience. Bone Marrow Transplantation (2010) 45, 1294–1299.

Related CTGA Records

External Links
https://ghr.nlm.nih.gov/condition/griscelli-syndrome

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