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BACKGROUND: This study examines gene-environment interaction between the *MTHFR* C667T polymorphism and folic acid in the etiology of orofacial clefts (OFC). We used a pooled-analytical approach on four studies that used similar methods. **METHODS:** We used logistic regression to analyze the pooled sample of 1149 isolated cases and 1161 controls. Fetal and maternal *MTHFR* C677T genotypes, and maternal periconceptional exposure to smoking, alcohol, vitamin containing folic acid and folic acid supplements were contrasted between the cleft types [non-syndromic clefts lip or without cleft palate (CL(P)) and non-syndromic cleft palate (CP)] and control groups. **RESULTS:** There was a reduced risk of CL(P) with maternal folic acid use



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This study selected individuals from a private practice who had cement-retained implant restorations and were scheduled for regular implant maintenance or were consulted because of a complication. Mechanical complications included all incidents (extensive porcelain chipping, framework fracture, abutment screw loosening) that required the removal of the restoration. Biological complications included peri-implantitis and peri-implant mucositis. Peri-implantitis was diagnosed if an implant had bleeding on probing, pocket depths 6 mm or more and progressive crestal bone loss exceeding 1.5 mm after first year of service (Fig. <u>1</u>a and b). Peri-implant mucositis was defined as a swelling, bleeding on probing, and increased probing depths of peri-implant tissues without evident progressive bone loss radiographically, which does not exceed acceptable norm, established by Albrektsson et al. (<u>1986</u>) (Fig. <u>2</u>a and b).



Figure 1. (a)Probing of an implant with peri-implantitis; (b) Radiographic evidence of progressive bone loss. ⊽ C

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Figure 2. (a) Bleeding and swelling of peri-implant tissues around implant restoration; (b) Radiographic image of peri-mucositis. Crestal bone loss does not exceed acceptable norms.

Radiographic images were taken with RVG Windows Trophy 5.0 (Trophy Radiologie Inc, Paris, France) using a paralleling technique with Rinn-like film holder in high-resolution mode. To define the extent of crestal bone loss, control radiographic images were compared to radiographs taken at the time of complication. To confirm diagnosis, peri-implant tissues were probed with 1.0 mm marked periodontal probe (Hu-Friedy, Chicago, IL, USA) and bleeding and suppuration (if present) were recorded.

In the case of mechanical complication, the implant restoration was removed by perforating occlusal/palatinal surfaces to gain access to the abutment screw. The retrieved abutment-restoration complex and the peri-implant tissues were inspected for excess cement (Fig. 3). If



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Long-term safety and efficacy of sustained eculizumab treatment in patients with paroxysmal nocturnal haemoglobinuria.

Hillmen P, Muus P, Röth A, Elebute MO, Risitano AM, Schrezenmeier H, Szer J, Browne P, Maciejewski JP, Schubert J, Urbano-Ispizua A, de Castro C, Soclé G, Brodsky RA.

St James's University Hospital, Leeds, UK. peter.hillmen@nhs.net

Abstract

Paroxysmal nocturnal haemoglobinuria (PNH) is characterized by chronic, uncontrolled complement activation resulting in elevated intravascular haemolysis and morbidities, including fatigue, dyspnoea, abdominal pain, pulmonary hypertension, thrombotic events (TEs) and chronic kidney disease (CKD). The long-term safety and efficacy of eculizumab, a humanized monoclonal antibody that inhibits terminal complement activation, was investigated in 195 patients over 66 months. Four patient deaths were reported, all unrelated to treatment, resulting in a 3-year survival estimate of 97.6%. All patients showed a reduction in lactate dehydrogenase levels, which was sustained over the course of treatment (median reduction of 86.9% at 36 months), reflecting inhibition of chronic haemolysis. TEs decreased by 81.8%, with 96.4% of patients remaining free of TEs. Patients also showed a time-dependent improvement in renal function: 93.1% of patients exhibited improvement or stabilization in CKD score at 36 months. Transfusion independence increased by 90.0% from baseline, with the number of red blood cell units transfused decreasing by 54.7%. Eculizumab was well tolerated, with no evidence of cumulative toxicity and a decreasing occurrence of adverse events over time. Eculizumab has a substantial impact on the symptoms and complications of PNH and results a significant improvement in patient survival.

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Long-term safety and efficacy of sustained eculizumab treatment in patients with paroxysmal nocturnal haemoglobinuria

Peter Hillmen,¹ Petra Muus,² Alexander Röth,³ Modupe O Elebute,⁴ Antonio M Risitano,⁵ Hubert Schrezenmeier,⁶ Jeffrey Szer,⁷ Paul Browne,⁸ Jaroslaw P Maciejewski,⁹ Jörg Schubert,¹⁰ Alvaro Urbano-Ispizua,¹¹ Carlos de Castro,¹² Gérard Socié,¹³ and Robert A Brodsky¹⁴

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Abstract

Paroxysmal nocturnal haemoglobinuria (PNH) is characterized by chronic, uncontrolled complement activation resulting in elevated intravascular haemolysis and morbidities, including fatigue, dyspnoea, abdominal pain, pulmonary hypertension, thrombotic events (TEs) and chronic kidney disease (CKD). The long-term safety and efficacy of eculizumab, a humanized monoclonal antibody that inhibits terminal complement activation, was investigated in 195 patients over 66 months. Four patient deaths were reported, all unrelated to treatment, resulting in a 3-year survival estimate of 97.6%. All patients showed a reduction in lactate dehydrogenase levels, which was sustained over the course of treatment (median reduction of 86.9% at 36 months), reflecting inhibition of chronic haemolysis. TEs decreased by 81.8%, with 96.4% of patients remaining free of TEs. Patients also showed a time-dependent

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Atypical combined immunodeficiency due to Artemis defect: A case presenting as hyperimmunoglobulin M syndrome and with LGLL

Bajin, T.Y.^a 💌 , Ayvaz, D.T.^b, Ünal, T.^c, Özgür, T.T.^b, Çetin, M.^c, Gümrük, F.^c, Tezcan, T.^b, de Villartay, J.-P.^d, Sanal, T.^b 👗

^a Department of Pediatrics, Hacettepe University Ihsan Doğramaci Childrens Hospital, Ankara, Turkey

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Abstract

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SCID can be caused by various genetic mutations leading to distinctive phenotypes according to the presence of T, B and NK cells. Artemis is a gene encoded on chromosome 10p. The deficiency of this molecule causes an inability to repair DNA double strand breaks and is one of the causes of radiosensitive T-B-NK+ SCID. The syndrome usually presents with opportunistic infections in the first years of life that leads to death if not treated with stem cell transplantation. The spectrum of the disease can be wide because of the heterogeneity of the mutations. Herein we present an atypical SCID (CID) patient with Artemis defect mimicking hyper IgM syndrome. Our patient had high serum IgM with low IgG and IgA levels, lymphocytosis and recurrent infections, intractable diarrhea, growth retardation, systemic CMV infection and sclerosing cholangitis. He also developed large granular lymphocytic leukemia and survived until the age of 6.5 years. © 2013 Elsevier Ltd.

Author keywords

Artemis; Hyper IgM syndrome; SCID

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Institution	Clinical Trials & Evaluation Unit, Royal Brompton Hospital & National Heart and Lung Institute, Imperial College, London, UK. Dipak.Kotecha@monash.edu	
Title	Erythropoietin as a treatment of <mark>anemia</mark> in heart failure: systematic review of randomized trials. [Review]	
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Scope Note for: Hyperthyroidism

MeSH HEADING: HYPERTHYROIDISM

SCOPE: Hypersecretion of THYROID HORMONES from the THYROID GLAND. Elevated levels of thyroid hormones increase BASAL METABOLIC RATE.

NOTE: THYROTOXICOSIS & THYROID CRISIS are available: do not make a diagnosis: use term of author

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▼ Filter By 1	measured at the thyroid gland, and effective dose (E) were assessed for both imaging modalities. The mean ESD measured at the thyroid gland was the highest at 120 kVp, followed by the 100 kVp DSCT and the ICA protocols with 4.0+/-1.8, 2.7+/-1.0 and 1.1+/-1.2 mGy, respectively. The mean E was estimated to be 10.3+/-2.1, 6.2+/-2.3 and 5.3+/-3.4 mSv corresponding to the 120-kVp, 100-kVp DSCT and ICA protocols, respectively. The application		
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Species	Latin America. This study retrospectively analyses the clinical characteristics and survival of 27 patients with MPAL evaluated in three medical institutions of Mexico. All cases meet World Health Organization 2008 criteria: 70.3 % of patients had B lumphoid/mulliol lineage MPAL induction chemotherapy protocols included.	STUDIES ON TRANSMISSIBLE LYMPHOID LEUCEMIA OF MICE. [J Exp Med. 1931]
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<u>Clear all</u>	induced aplasia during remission induction (5.2 %). In 68 % of cases, we were able to administer maintenance therapy as a regimen in lymphoblastic leukemia. At the time of analysis, 70.4 % of the patients in the entire	See more
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