

Chronic Granulomatous Disease (CGD): a European database

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CGD is an immunodeficiency caused by deletions or mutations in genes that encode subunits of the NADPH oxidase complex. Normally, assembly of the NADPH oxidase complex in phagosomes of phagocytic cells leads to a "respiratory burst", essential for the clearance of phagocytosed microorganisms. CGD patients lack this mechanism which leads to life-threatening infections and granuloma formation.

However, a clear picture of the clinical course of CGD is hampered by its low prevalence (~1:250.000). Therefore, extensive clinical data from 438 European patients were collected.

Of these patients, 359 males and 79 females, X-linked CGD (gp91) accounted for 52 % of the cases, autosomal recessive inheritance for 29 % whereas in 19 % no subtype was established. At the time of inclusion 60 % of the patients were alive, 21% deceased and 19 % lost to follow up.

The disease manifested itself most frequently in the lungs (64% of patients), lymph nodes (50%), skin (50%), gastrointestinal tract (49%) and liver (32%). The most frequently cultured microorganisms per episode were *S. aureus* (30%), *Aspergillus* (26%), and *Salmonella* spp. (16%). Surprisingly, *Pseudomonas* (2%) and *Burkholderia* (<1%) spp. were found only sporadically.

71% received antibiotic maintenance therapy, 53% antifungal therapy. 33% were treated with gamma-interferon.

Most prominent reason of death was pneumonia/pulmonary abscess (18/91 cases), septicaemia (16/91) and brain abscess (4/91). 24 patients (5%) received a stem cell transplantation.

These data provide further insight in the clinical course of CGD and could be of help in optimizing the treatment of these patients.