Zeidler et al. Orphanet Journal of Rare Diseases 2010, 5(Suppl 1):P22
http://www.ojrd.com/content/5/S1/P22

ORPHANET JOURNAL OF RARE DISEASES

POSTER PRESENTATION Open Access

International registry: genetic and phenotypic characteristics of a heterogenous group of disorders

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From 5th European Conference on Rare Diseases (ECRD 2010)
Krakow, Poland. 13-15 May 2010

Severe congenital neutropenia (CN) stands for a group of disorders characterised by extremely low neutrophil counts (ANC < 0.5x109), early stage maturation arrest of myelopoiesis and recurrent bacterial infections. In general more than 90% of CN patients respond to daily G-CSF treatment with a sustained neutrophil increase resulting in significantly reduced infections and an improved quality of life. Besides neutropaenia, the differences in treatment response and the presence of various concomitant clinical features in subpopulations of patients in conjunction with an increased risk of leukaemia transformation in about 10% of all CN patients strongly suggested to search for new sub diagnoses to identify patients at risk of leukaemia.

Within Europe the SCNIR has collected longitudinal clinical data on more than 493 patients with various causes of CN (289 congenital, 64 cyclic, 132 idiopathic and 8 others) from 22 countries. This unique resource of data was used to identify new genes, classify patients by genetic subtypes of CN, estimate their relative frequency and correlate genetic subtypes with prognosis and outcome.

To date, more than 10 disease causing gene mutations have be identified in congenital neutropaenia patients. In approximately 50-60% of all patients autosomal dominant mutations in the ELANE gene are present. Initial genotype-phenotype correlation identified a group of different genetic defects sharing a high risk of leukaemia transformation in contrast to others with no increased risk of leukaemia.

The identification of new CN subtypes, their distinctive risk of malignant transformation and the response to treatment has contributed substantially to our general understanding of neutropaenia. New risk adapted strategies for diagnosis and treatment have to be implemented in the management of CN patients.

Published: 19 October 2010

doi:10.1186/1750-1172-5-S1-P22
Cite this article as: Zeidler *et al.*: International registry: genetic and phenotypic characteristics of a heterogenous group of disorders. *Orphanet Journal of Rare Diseases* 2010 **5**(Suppl 1):P22.

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