

## Whole genome sequencing of NK leukaemia

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**Introduction:** Natural killer (NK)-cell leukaemia is common in Asia and South America populations. This is a fatal disease with the patients' median survival time of around 2 months. Little is known about the molecular characteristics and pathogenetic mechanisms of the illness. The aim of this study was to identify novel pathogenic mutations in NK bone marrow by whole genome sequencing.

**Methods:** An adult NK leukaemia patient bone marrow sample sorted with CD56+ together with the skin biopsy were captured for whole genome sequencing. The somatic sequence changes identified will be further validated in other NK bone marrow samples by Sanger sequencing.

**Results:** The whole genome in both samples has an average coverage at 40X. A total of 42 single-nucleotide variations (SNVs) were identified in the NK bone marrow sample but absent in the skin biopsy. However, no recurrent SNVs were identified in three other NK bone marrow samples.

**Conclusion:** The identified SNVs need to be further validated with an increased number of NK bone marrow samples.

## The psychosocial burden of psoriasis and barriers to biologic therapy in Hong Kong: patients' perspectives

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**Introduction:** Psoriatic skin lesions are often visible and have negative impact on patients' quality of life (QoL). The introduction of biologic therapies has revolutionised the treatment of psoriasis. They are highly efficacious in clearing skin lesions and relieving joint symptoms. The use of biologics in psoriasis patients is associated with improved QoL and patient satisfaction. In Hong Kong, the psychosocial impacts of psoriasis on patients and their satisfaction towards various treatment modalities have not been well studied. Moreover, the ways local patients come to access the information on biologics and their concerns are unclear.

**Methods:** We conducted a survey of 85 psoriasis patients from a patient self-help group to discern the disease impacts on QoL and patient attitudes towards biologics. The patients' self-perceived disease severity and the disease impacts on physical, psychological, and social well-beings were assessed by a 10-point scale (1=minimal; 10=maximal).

**Results:** The results show that 29% of patients (n=25) had mild (body surface area [BSA] <3%), 36% (n=31) had moderate (BSA 3-10%), and 34% (n=29) had severe (BSA >10%) psoriasis. The male-to-female ratio was 1.4:1. The mean age was 52.6±13.6 years. The median duration of disease was 8 years (3-13 years). The self-perceived disease severity was 7.68±2.70. Patients with moderate-to-severe psoriasis was associated with significantly higher self-perceived disease severity (P<0.01). The perceived disease impacts on six thematic issues were analysed: emotion (7.36±2.74); social life (6.92±2.81); general physical health (6.76±2.76); economy (5.67±3.20); job opportunity and work (5.61±3.30); and family life (4.76±2.64). Nearly half (48%) of patients was unsatisfied with current treatments. The main reasons were lack of treatment efficacy (58%), side-effects (29%), and high cost (20%). Only six (7%) patients had received biologic therapies; 46% of patients had never been informed or aware of biologic treatments in psoriasis. Most patients learned biologics through media (78%), medical personnel (20%), self-help groups (13%), and internet (11%).

**Conclusion:** Psoriasis has negative impacts on patients' QoL, especially on psychosocial aspects. Majority of patients were unsatisfied with current treatments due to suboptimal disease control. Biologic therapy is underutilised in management of skin psoriasis and there is a lack of public awareness of such treatment modality. Medical professional has a potential pivot role in patient education of biologic therapy.