

# Multidisciplinary Care for Adult Hermansky-Pudlak Syndrome Patients in Puerto Rico

Isabel Becerra<sup>1</sup>, Shirley Martinez<sup>1</sup>, Atencio Xavier<sup>2</sup>, Torrens Jonathan<sup>2</sup>, Juan Alemany<sup>3</sup>, Eric Cruz<sup>3</sup>, Felix Rivera Borges<sup>3</sup>, Monica Egozcue<sup>3</sup>, Jesse Roman<sup>4</sup>, Rosa Roman<sup>2,3</sup>

<sup>1</sup>Universidad Autonoma de Guadalajara, Guadalajara, Mexico; <sup>2</sup>Ponce Health Sciences University, Ponce School of Medicine, Ponce, Puerto Rico; <sup>3</sup>Mayaguez Medical Center, Mayaguez, Puerto Rico; <sup>4</sup>Jane & Leonard Korman Respiratory Institute, Thomas Jefferson University, Philadelphia, PA

## Abstract

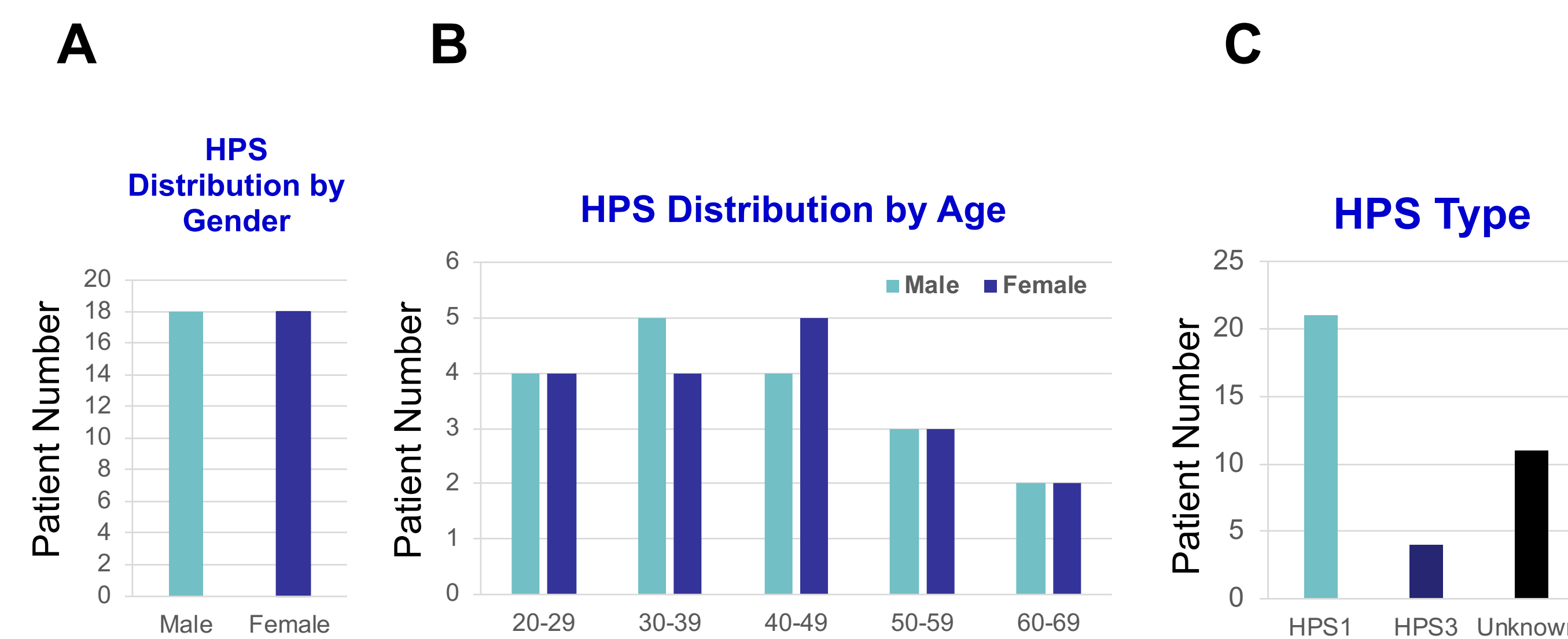
**Background:** Hermansky-Pudlak syndrome (HPS) is an autosomal recessive rare disease characterized by oculo-cutaneous albinism, bleeding diathesis due to platelet dysfunction, and is sometimes accompanied by immunodeficiency, granulomatous colitis, and/or interstitial lung disease / pulmonary fibrosis. Puerto Rico houses the highest concentration of patients with HPS, especially HPS-1, which is considered quite severe and often associated with pulmonary fibrosis. We believe that the establishment of an HPS Multidisciplinary Clinic in Puerto Rico would improve outcomes.

**Methods:** A multidisciplinary clinic was started in November 2018 and another was held in February 2019 at the Mayaguez Medical Center located in the Southwest region of the island. Pulmonologists, hematologists as well as gastroenterologists and a dentist staffed the clinic. Pulmonary function testing was available. Patients were identified by the HPS Network and other announcements.

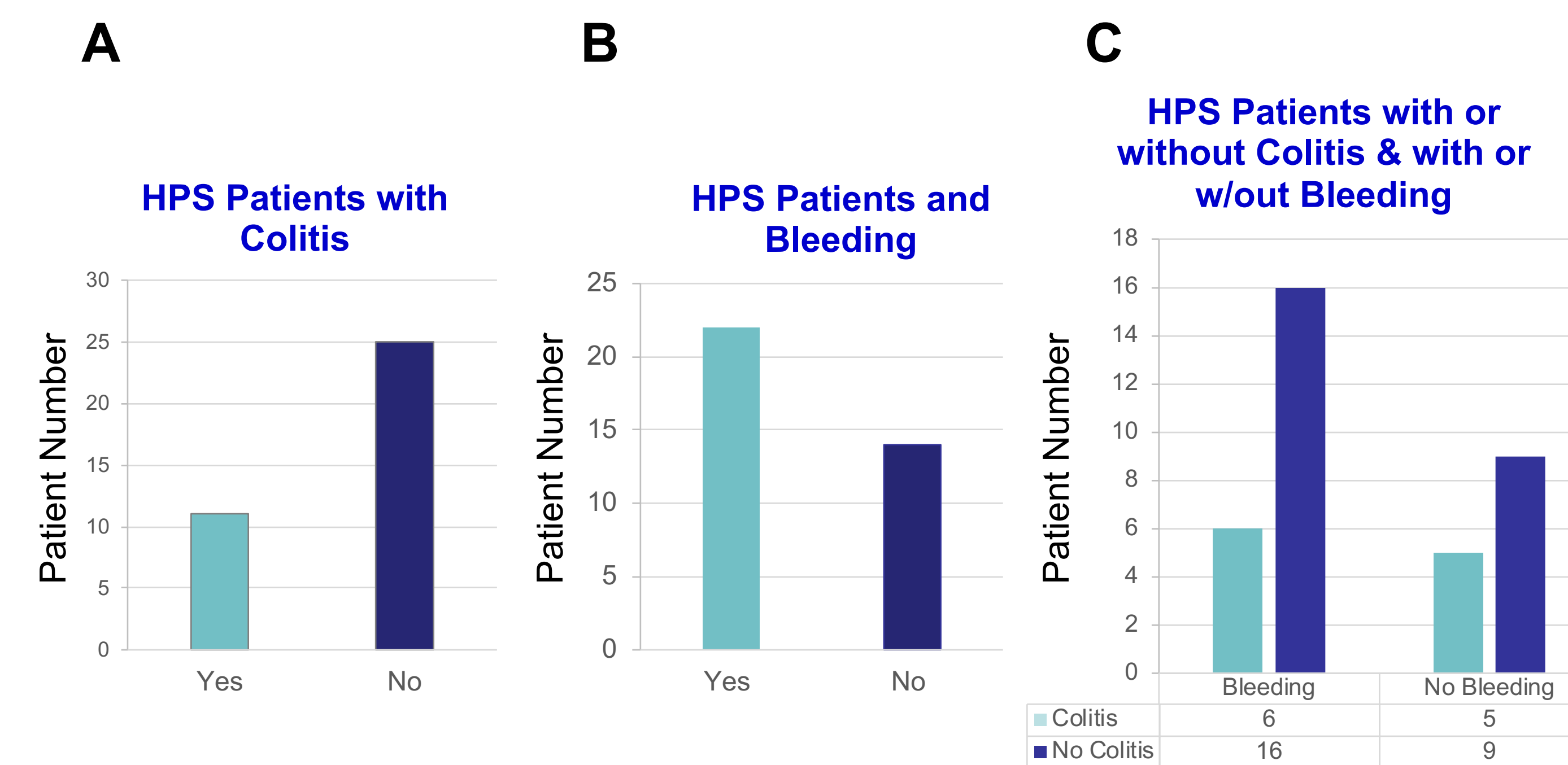
**Results:** Thirty-six patients were evaluated during the February 2019 clinic. Gender distribution was essentially equal and most patients were 50 years of age or younger. Fifty-eight percent classified themselves as having HPS-1 and 11% as having HPS-3, but the exact mutation was not known in 31%. Of the thirty patients obtaining pulmonary function tests, 60% showed abnormal force vital capacity (FVC) with 13% showing severe dysfunction. Thirty-one percent of patients had been treated for colitis, while 58% had bleeding at some point. Most patients had not received Influenza vaccination.

## Results

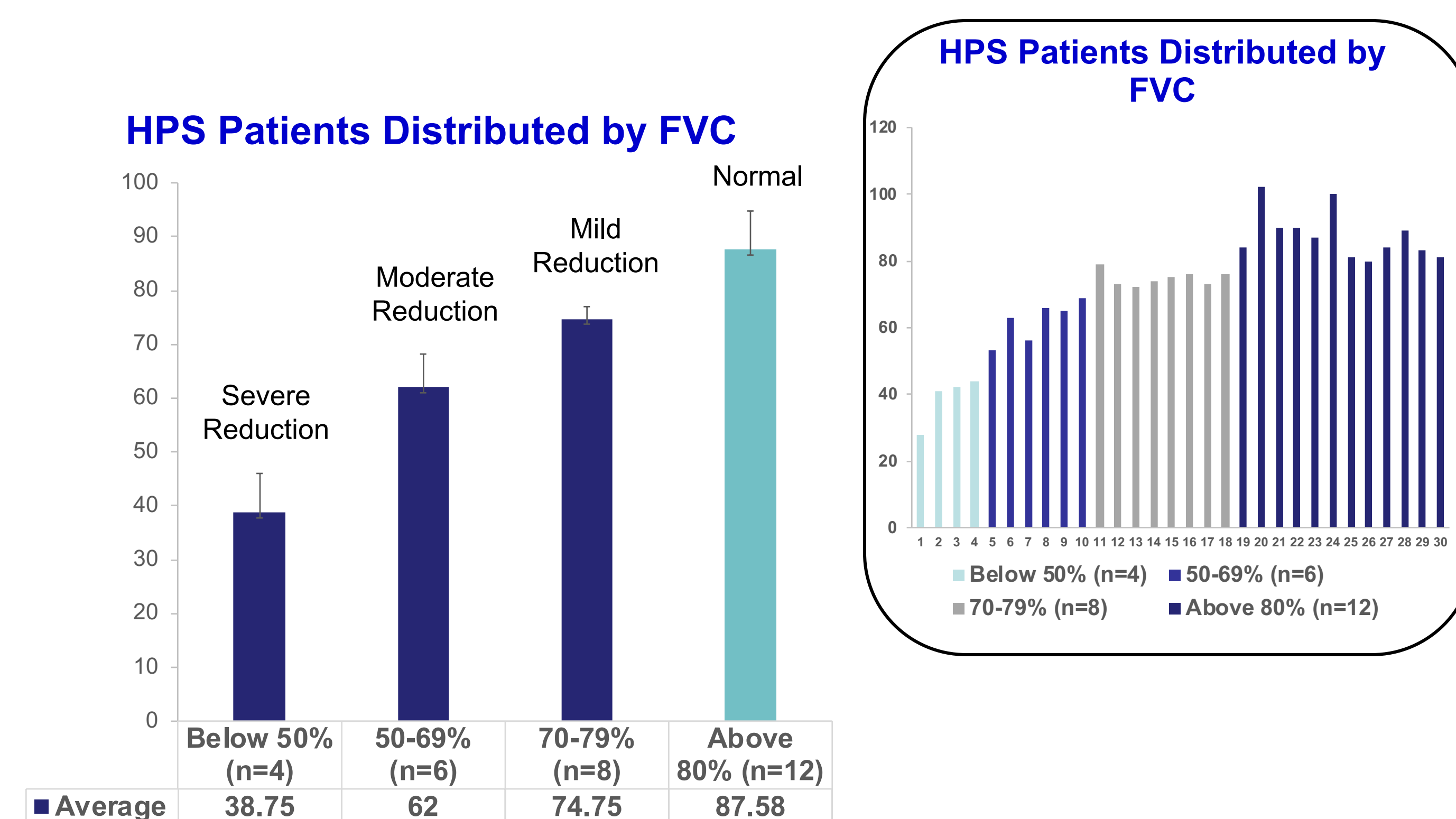
### Demographics



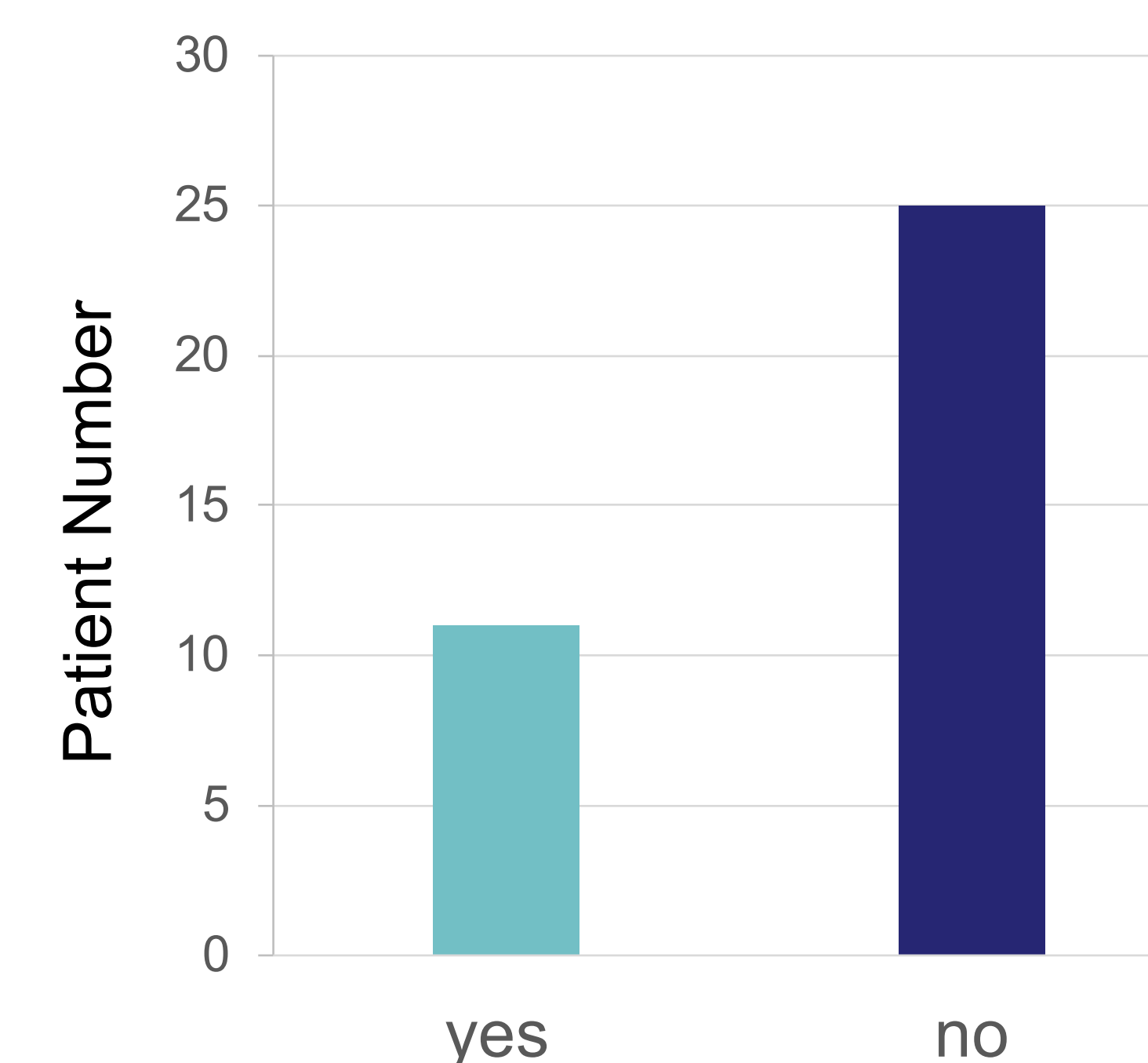
### Prevalence of colitis and/or bleeding disorder



### Most patients had lung dysfunction



### HPS patients with Influenza vaccine



## Conclusions

1. Most patients attending the clinic had significant manifestations of the disease including pulmonary dysfunction, colitis and bleeding.
2. The majority had HPS-1, but the exact mutation was not known in one third of cases.
3. The preventive care and management of HPS and its complications is complex and might be better delivered via a multidisciplinary clinic that could serve not only as a clinical home for patients, but also as resource for information and clinical trials.

## Acknowledgements

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