Hermansky-Pudlak syndrome (HPS) is a rare autosomal recessive disorder consisting of a triad of oculocutaneous albinism, platelet dysfunction and accumulation of ceroid-like depositions in tissues [1]. We report five patients with HPS which have various clinical symptoms associated with gastrointestinal complications related to chronic granulomatous colitis.

Case 1: 28-year-old female patient whose HPS was diagnosed in 2001. Performed colonoscopy showed mucosal ulcers, pseudopolyps and biopsy results showed granulomatous colitis.

Case 2: 28-year-old male patient whose HPS was diagnosed in 2009. Performed colonoscopy showed edema from anal canal up to the cecum, biopsy results showed granulomatous colitis.

Case 3: 28-year-old male patient whose HPS was diagnosed in 2011. Colonoscopy doesn’t perform yet, but clinical symptoms were compatible with Chron’s Disease.

Case 4: 32-year-old female patient whose HPS diagnosed in 2013. Performed colonoscopy showed compatible results with ulcerative colitis with typical distribution of inflammatory regions, biopsy results showed inflammatory bowel disease.

Case 5: 33-year-old female patient whose HPS diagnosed in 2013 is the sister of case 1. Colonoscopy doesn’t performed yet. There are no signs of inflammatory bowel disease. Each case of platelets have increased spreaded glycogen and plenty of dense bodies. Even though in case 4 glycogen stores and alpha granules were seen. In case 3 and case 5, the platelets size were larger than the others. Each case of platelets were seen irregular in shape. The platelets ultrastructural examinations will keep going by applying various aggregating agents (fibrinogen, collogen, adenosine diphosphate, epinephrine) to the C-PRP of the patients which are treating with various medicines.

References
Fig. 1: DB (dense bodies), AG (alpha granules), grey arrow (glycogen), G (glycogen store), OCS (open canalicular system), black arrows: a canaliculus associated with outside.