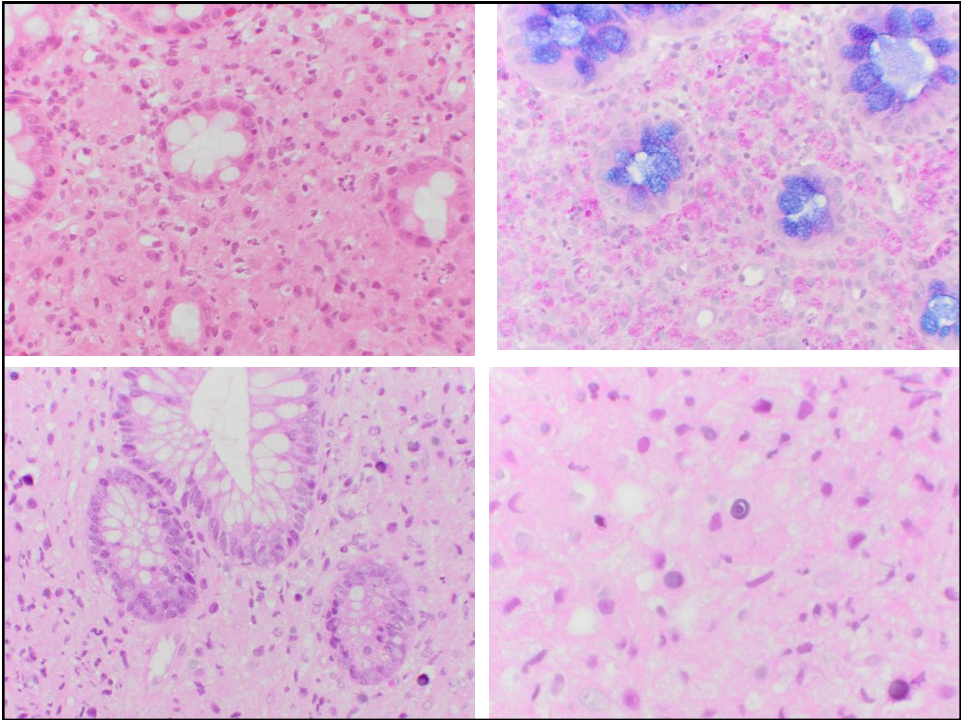
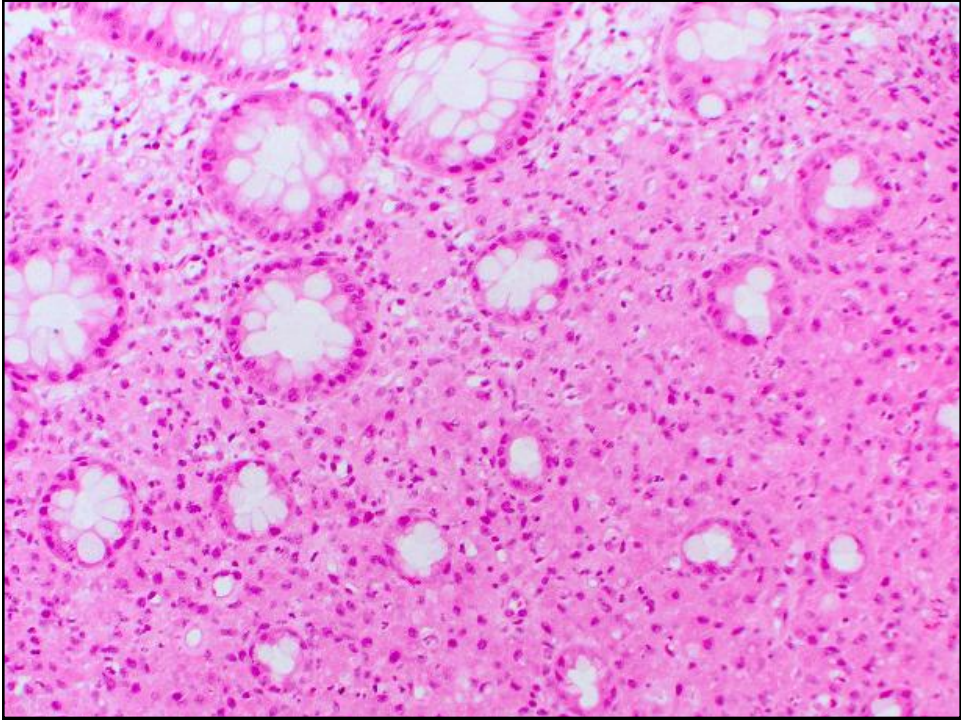
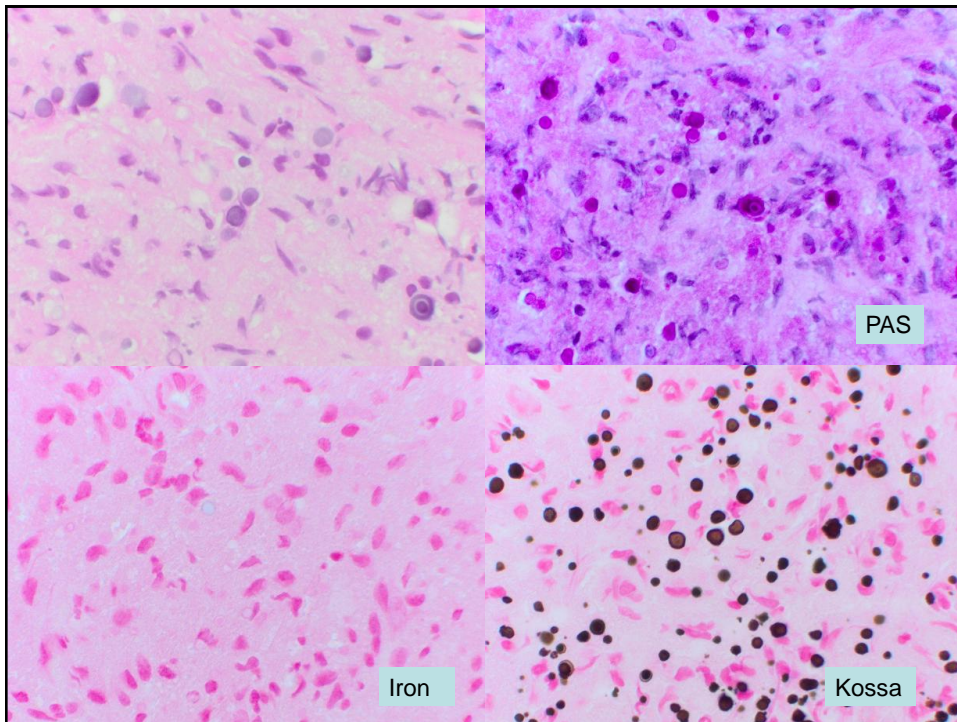


Eleven months old girl

- Borne with „CHARGE syndrome“
- including coloboma, choanal atresia, deafness, palsy of the left 7th and 8th cranial nervs and right-sided aortic arch

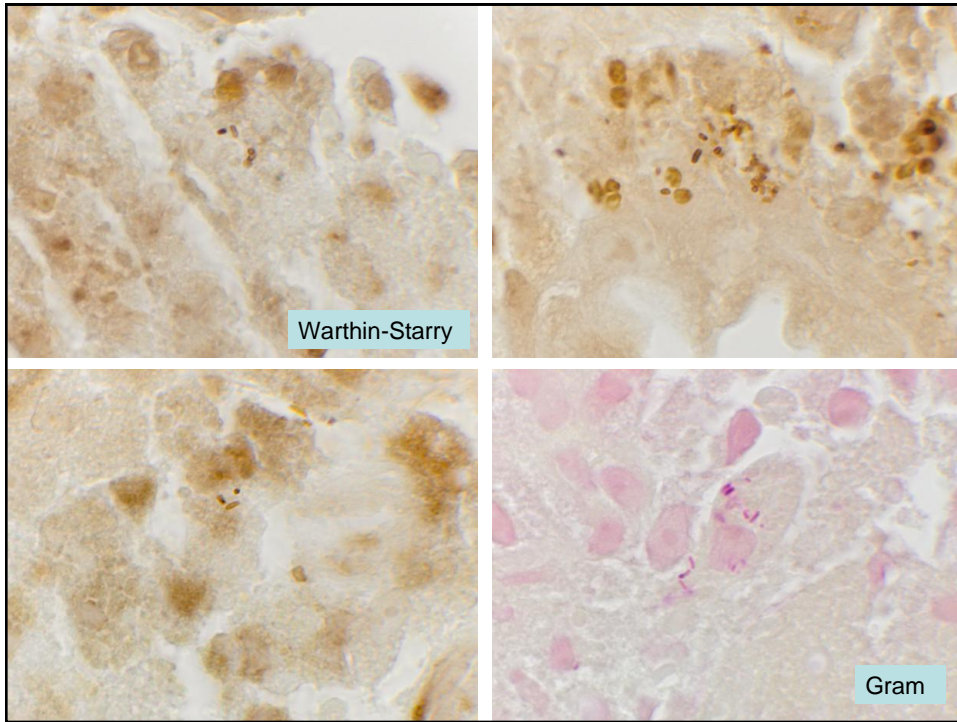
- Recurrent airway infections and recurrent hemorrhagic diarrhea
- Endoscopy at 11 months: Multiple plaque-like colonic ulcers, predominantly in the right colon



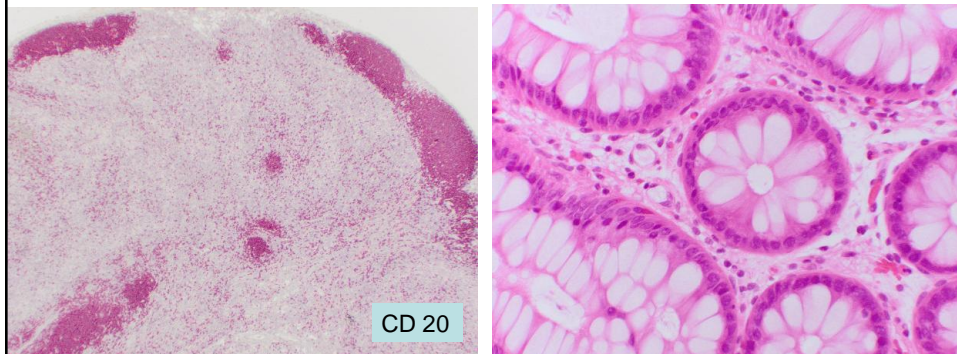


Malakoplakia

- Unusual tissue response to bacterial infection
- characterized by von Hansemann cells and Michaelis-Gutmann bodies
- indicating immune deficiency interfering with normal phagocytic degradation
- E.coli, Aerobacter, Klebsiella spp, Rhodococcus spp, Yersinia, Mycobacteria, fungal infection



- Immune status:
- Lymphopenia (>CD4 positive T-cells) and hypogammaglobulinemia
- Severe combined immunodeficiency (SCID)



„CHARGE syndrome“

- Malformational syndrome involving among other structures the third and fourth pharyngeal pouch
- Altered morphogenesis of thymus? (no autopsy)

- Charge syndrome
- Including thymic hypoplasia (?) B-cell and macrophage phago-lysosome-defect
- Recurrent intestinal bacterial infections
- Malakoplakia

Follow-up

- Progressive decline in T and B cell function, recurrent bacterial airway infections and terminal respiratory failure at the age of eighteen months