Pediatric Emergency Department(Out-patient)

2019-05-28

Chief complaint: right lower abdomen pain for 2 days.

Current medical history: No apparent cause pain, paroxysmal pain, no fever, no nausea, no

vomiting, no diarrhea.

Physical examination: Mild tenderness in the right lower abdomen.

Assistant examination: Abdominal ultrasound revealed no abnormal change.

Diagnosis: Abdominal pain Treatment: Oral probiotics.

Pediatric General Surgery Department(Out-patient)

2019-06-20

Chief complaint: Conservative treatment of right inguinal inflammation for half a month.

Physical examination: Bilateral Inguinal area did not touch the mass.

Assistant examination: The ultrasound revealed no uterus and solid nodules in the bilateral inguinal region.

Pediatric Urology Department (Hospitalization)

2019-09-27

Chief complaint: Examination revealed absence of uterus for 4 months.

Physical examination: External appearance of undeveloped female genitalia without evident labia majora and labia minora. No pubic hair, no clitoral hypertrophy, no scrotal or testicular tissue. Vaginal opening and urethral opening are visible. Bilateral Inguinal area did not touch the mass.

Assistant examination: high levels of LH and FSH. Decreased Inhibin-B.

Treatment: Psychological consultation: psychological sex was female. The patient was operated after DSD multidisciplinary team consultation and the consent of parents.

Pediatric General Surgery Department(Hospitalization)

2019-05-30

Chief complaint: pain in right inguinal region for 3 days.

Current medical history: no relief in the abdominal pain after oral probiotics. no fever, no nausea, no vomiting, no diarrhea.

Physical examination: Local redness and swelling of the right inguinal region (5.0*2.5cm), tenderness, no wave sensation, without clear boundary, no adhesion to the epidermis. Left inguinal region was normal.

Assistant examination: WBC: 11.92x10°/L, Neutrophil %: 60.8%, CRP: 54.50mg/L. The ultrasound revealed bilateral inguinal mass and round ligaments with terminal inflammation are more likely.

Diagnosis: Right inguinal cellulitis

Treatment: Anti-inflammatory symptomatic treatment, local redness and swelling vanish gradually, pain relived.

Pediatric Urology Department (Out-patient)

2019-06-28

Chief complaint: Examination revealed absence of uterus for 1 week.

Physical examination: External appearance of female genitalia without evident labia majora and labia minora. No clitoral hypertrophy, no scrotal or testicular tissue. Vaginal opening and urethral opening are visible.

Assistant examination: high levels of LH and FSH. The karyotype was 46,XY and positive SRY gene. The results of three-day HCG-stimulation test was negative as well as seven-day HMG-stimulation test. Targeted next generation sequencing revealed that NR5A1 gene mutation was detected in the patient.

Diagnosis: 46 XY DSD Treatment: Hospitalization

Pediatric Endocrinology Department (Hospitalization)

2020-03-15

Chief complaint: Examination revealed Gonadal dysplasia for 10 months.

Physical examination: External appearance of undeveloped female genitalia without evident labia majora and labia minora.

Assistant examination: high levels of LH and FSH. Decreased Inhibin-B and AMH.

Treatment: Low-dose estrogen replacement therapy.

Supplementary Fig. 1. Timeline of the diagnostic process for the patient.