This manuscript describes an extremely rare disease, the diagnostic process, and the course of this disease. The main point however is the role and the importance of genetic and prenatal diagnosis when physicians have to deal with rare genetic diseases, and genetics and prenatal diagnosis can change the management of these conditions.

Page(P) 1 Line (L) 2, the incidence of the disease and the number of cases described so far are missing in the text and should be included especially in the case of rare diseases.

Answer: The precise prevalence of UCMD is not sufficiently known, with an estimated 0.13 per 100,000 in Northern England, which is much higher than what is reported in China.

P1 L 24 a more detailed patient history should be included. At birth, did the brothers already have signs of the disease? When did the parents and the physician apprise the first signs and symptoms of the disease?

Answer: Prenatal movements might be reduced in fetuses with UCMD. Some patients have congenital hip dislocation, torticollis and transient kyphotic deformity. Some transient feeding difficulties or poor sucking might occur in the neonatal period. The most common presentations are delayed motor milestone and proximal muscle weakness.

The changes in the brothers' condition, such as feeding problems and hip dislocation, were not be noticed for their parents' poor medical knowledge. The brothers were admitted to our hospital for the first time because of their disabilities in walking independently. Therefore, it is difficult to provide more detailed medical history data.

P1 L29 simple pregnancy should be changed into uneventful.

Answer: The first child of the mother was given birth by cesarean section for the incorrect fetal position. The second child was delivered directly by cesarean section for the scarred uterus.

P2 L 45 were these mutations novel or were already present in the common databases?

Answer: The two mutations were already present in the common databases.

P2 L 73. Some information about the pregnancy is missing; was the second-trimester ultrasound normal? Was there any fetal sign that could be apprised like few fetal active movements? Why amniocentesis was preferred over Chorionic villus sampling?

Answer: There were not routine prenatal check-ups for the fetus. It was not known whether the second-trimester ultrasound of the fetus was normal. The mother felt that the fetal movements were normal since the 16th week of pregnancy.

Amniocentesis is usually at 16-22 weeks of gestation. 10-20ml of amniotic fluid containing cells shed is extracted from the uterus to check for the diseases by the ultrasound guidance.

Chorionic villus sampling is usually in the first trimester of gestation. A small number of fetal villi containing fetal cells in the outer trophoblast are extracted from the uterus to check for the diseases by ultrasound guidance.

The fetus had been pregnant for 20+6 weeks and was suitable for amniocentesis examination.

P3L96 Some information about the pathogenesis of the disease should be included.

Answer: Variable degrees of histological changes can be observed in muscle biopsies of patients with UCMD. The spectrum includes fiber size variation, increased endomysial connective tissue or adipose tissue, and mild necrotic and regenerating process. Collagen VI staining in muscle biopsies of patients with UCMD is variably less or full absent in the extracellular matrix. It is present in the interstitium but is absent or reduced in the sarcolemma.

L3 l 78 Was an autopsy performed? Was the genetic test performed also after birth?

Answer: The parents refused to do an autopsy and genetic test for the third child after birth.

P4 L 83 and P4 L 134 and in the whole text "prevented the birth of" should be changed into opted for voluntary interruption of pregnancy

Answer: The words of "prevented the birth of" is changed into "opted for voluntary interruption of pregnancy".