P05.05. Prenatal presentation of fibrochondrogenesis: a rare lethal skeletal dysplasia. Fiona H. Langdon¹, Jan E. Dickinson¹,², Fiona McKenzie¹, Andrew Gill¹,²
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Case
A 31 year old woman at 20 weeks gestation was referred following a mid-trimester morphology ultrasound suggestive of a fetal skeletal dysplasia. On review, the fetal long bones all measured below the first centile but exhibited normal mineralization. The long bones were significantly curved & had flared speckled metaphyses, particularly of the humeri & femora. The tubular bones of the hands & feet were unaffected. The hands were malpositioned with the thumb & index finger apposed & the lateral three digits closely approximated. The ribs were short & the thorax small. Platypondyly of the spine with poor mineralization of the posterior vertebral elements was evident. There was midface hypoplasia, frontal bossing & brachycephaly. Chromosomal microarray was normal & FGFR gene mutation studies were negative. The parents declined pregnancy termination. Interval fetal growth was suboptimal & amniotic fluid volume increased progressively to that of polyhydramnios. Delivery occurred via elective repeat caesarean section at 38 weeks gestation of a 2360g female.

Prenatally a decision to not perform invasive ventilation had been made, in consultation with the parents. The neonate died at 18 days of life from respiratory failure. Postmortem skeletal survey indicated a diagnosis of fibrochondrogenesis, however gene mutation studies were negative for COL11A1 & A2 genes, but positive for TRPV4; altering the final diagnosis to severe lethal metatropic dysplasia.

Conclusion
Molecular genetic testing now provides potential for accurate prenatal & postnatal diagnosis of rare skeletal dysplasias. As in this case, gene mutation analysis of the proband & parents substantially altered the recurrence counselling from 25% recurrence (fibrochondrogenesis) to sporadic (lethal metatropic dysplasia).