Introduction
The 3MC syndrome is a unifying entity for Mingarelli, Malpuech, Michels and Carnevale syndromes. Initially believed to be separate genetic disorders, these four syndromes are now known as 3MC syndrome collectively. Affected individuals have a distinctive facial gestalt with high-arched eyebrows, ptosis and hypertelorism. Features such as orofacial clefting, umbilical abnormalities, ocular and skeletal defects can be variably present. Intelligence of the affected individuals can be normal. It is a rare autosomal recessive genetic disorder related to MASP1, COLEC11 or COLEC10 gene mutations. Mutation in these genes have detrimental effect in cell migration control in early embryonal development. To date, less than 50 molecularly confirmed individuals were reported worldwide.

Case report
The proband was a 20 years old Pakistani gentleman born at term after an uncomplicated pregnancy and delivery. Parents were consanguineous and phenotypically normal. He had history of bilateral cleft lip and palate, omphalocele, left undescended testis and hypoplastic left ectopic kidney with impaired perfusion and function. He also had bilateral serous otitis media with myringotomy performed and required hearing aids. Intellectual development was normal. On physical examination, he had dysmorphic features including hypertelorism, ptosis and highly arched eyebrows. (Fig.1) The proband’s youngest sister, who was 8 years old, was found to have a similar clinical phenotype. She had an atrial septal defect presented with heart murmur and Chiari I malformation detected by MRI brain. She had normal intellectual development otherwise. Physical examination revealed similar dysmorphic features and supraumbilical depression. (Fig. 2) Medical exome sequencing was performed on DNA extracted from peripheral blood revealed both sib-pair harboured homozygous pathogenic NM_139125.3 [MASP1]:c. 1993C>A (p.Gly665Ser) [PM2, PM5, PP1, PP3, and PP4]. Parents were asymptomatic heterozygous carriers. This variant is classified as likely pathogenic according to the ACMG guideline. The molecular diagnosis of 3MC syndrome was substantiated.

Conclusions
In this case report we wish to illustrate a sib-pair with MASP1 related 3MC syndrome born to consanguineous parents. This is the first reported family with molecularly confirmed 3MC syndrome in Hong Kong.

Fig 1. Clinical photo of the proband showing features including hypertelorism, blepharoptosis and high arched eyebrows.
Fig 2a,b Clinical photo of the proband’s youngest sister showing similar facial features and supraumbilical depression.