RECURRENT TRISOMY IN SUCCESSIVE PREGNANCIES

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A 28yr lady in nonconsanguinous marriage with no relevant medical surgical or family history had 4 pregnancies successively at intervals of 2 yrs.each.1st pregnancy with routine antenatal checkup,no detectable anomalies on USG,First Trimester Screening not done,ended up in LSCS for PROM at 36wk delivering live boy baby weighing 3.25KG,suspected at 3 mths due to delayed milestones, abnormal facies,diagnosed as Downs Syndrome with KT 47xy+21,Parental KT normal.Her 2nd pregnancy ended in a miscarriage at 8wk,D&C sample unsuitable for cytogenetic study.Her 3rd pregnancy at 12-13wk NT 2.6mm,age related risk 1:210 for T21,by NT1:40,amniocentesis at 16wk,KT 47+21,terminated at 19wk,dead male fetus,patient refused autopsy.Her 4th pregnancy at 11wk NT 2.4mm hypoplastic nasal bone,subjected to Chorionic Villus Biopsy,KT 47+21, terminated,refused autopsy.Patient did not consent for any further evaluation and lost for follow up.

Conclusions.T21 could be due to Meiotic Nondisjunction,Cryptic Parental Gonadal Mosaicism,Robertsonian Translocation usually with chromosome14,Duplication Here recurrence of T21 could be due to parental gonadal Mosaicism which could have been evaluated if patient had agreed.