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Mosaic trisomy 22 in a girl with severe bone deformity

Anum Fatima[1], Mohsina Ibrahim[2]
[1] Child Aid Association, Pakistan
[2] National Institute of child health, Pakistan

Abstract
Mosaic Trisomy 22 with minimal physical findings and normal development are under diagnosed we report here a case of mosaic trisomy 22 i.e. 46, XX/47, XX, +22 in a girl aged 16 years. Most of the time trisomy 22 is seen in abortus material, In contrast live birth of MT22 is rarely seen due to severe organ malformations and bone deformities were visible after age of 1 year. The boney deformities included swelling in feet, rigidity in hands and legs. The patient also had congenital heart disease (Atrial Septal Defect).Cytogenetic analysis was carried out on peripheral blood lymphocytes, twenty metaphases were analyzed and revealed with mosaic trisomy of chromosome 22. The severity of the disorder can depend on the number of cells in which the extra chromosome 22 is present.

Keywords: trisomy, Girl, deformity, physical

Introduction
Mosaic trisomy 22 was first described by (Schinzel in 1981). Since then, there have been about 20 reports on live born children with mosaic trisomy 22 (MT22). It is speculated that children with mosaic trisomy 22 with minimal physical findings and normal development are under diagnosed. The condition appears to occur in females more frequently than in males (approximately 3 females: 2 males) (NORD National Organization of Rare Disorder).
The presence of an additional G-banding chromosomes in the dificiency of slowness has been reportable in some case (for reviewer see Hsu *et al*, 1971 and Gustavsn *et al*, 1972).The use of Giemsa techniques in recent studies enabled the diagnose of chromosomal abnormality twenty two to be created in rare case (Bass *et al*; Zackai *et al*,) [7] but not in others (Gustavson *et al*, 1972) with alike phenotypes.
We report here a case of mosaic trisomy 22 i.e. 46, XX (11), 47, XX, +22 (09) in a girl aged 16 years. Most of the time trisomy 22 is seen in abortus material. In contrast live birth of MT22 is rarely seen due to severe organ malformations. Chromosomal Abnormality represent a major cause of spontaneous abortion during the first trimester (Hassold *et al*;1980; Warburton *et al*; 1991) [4] and trisomy 22 has been identified as the third most common trisomy in non-conceiving cases representing 11-16% of all abortions due to chromosomal aberrations (ford *et al*; 1996, Menasha *et al*;2005). The severe organ malformations include microcephaly, cranial abnormalities, congenital heart disease, renal malformations and intrauterine growth retardation (IUGR). We report a case of young girl MT 22 with severe bone deformities and physical disability.

Patient Presentation
According to patient history, the girl has boney deformities with short stature (height 50 inches & weight 20kg). Boney deformities were first noted at age of 9 years due to theses deformities and severe disability the girl was investigated for congenital disorder. Her parents were first cousins and had unremarkable family history. But mother had the history of three still births. Mother was affected by polio virus, treated and operated upon leg deformity at the age of 9 years. Father history of drug addiction (heroin). Paternal age at conception was 35 years for the mother and 38 years for the father. All laboratory investigations were within normal range. Physical examination revealed undeveloped breasts and absent pubic hairs. She had no history of menstruation (primary amenorrhea). She had slight webbing of neck (image-1). Bone deformities were visible after age of 1 year. The boney deformities included swelling in feet, rigidity in hands and legs (image2, 3). The patient also had congenital heart disease (Atrial Septal Defect). Intellectually she was normal. Cytogenetic analysis was carried out on peripheral blood lymphocytes, twenty metaphases were analyzed and revealed with mosaic trisomy of chromosome 22 (image-4).

Discussion
In table-1 symptoms and signs of our patient are compared with those reported in literature with trisomy 22 (Hsuet *et al*; 1971; zellweger *et al*; 1975). Her phenotypic appearance is similar facial features reported MT22 girl earlier (Penchaszadeh & Coco 1975) [6]. In our patient the parents were first cousin but have normal phenotypic expression. This is the first report of a MT22 syndrome with a 46, XX /47XX +22 from Pakistan.
The mosaicism of our patient may be responsible for the lack of some important symptoms of the trisomy 22. Mosaic trisomy 22 is a rare chromosome disorder in which chromosome 22 is present three times, instead of the usual

two times, in some cells of the body. The severity of the disorder can depend on the number of cells in which the extra chromosome 22 is present.

Clinical features reported in Trisomy 22

- Microcephaly Abnormal ears
- Webbed neck Cardiac abnormalities
- Long fingers Growth retardation
- Renal malformation (missing, extra or underdeveloped kidneys)
- Shortened Limbs Mental Delay
- Hemidystrophy (abnormal development of each side of the body)
- Drooping eyelids Elbow malformations
- Abnormal or missing finger/toe nails
- Absent ovaries /fallopian tubes Undeveloped testes or ovaries (Penchaszadeh and Coco (1975) [6]

Table 1: Clinical features compare with patient history

Clinical features of trisomy 22 syndrome*	Patient case
Mental retardation	-
Growth retardation	+
Microcephaly	-
Cardiac Abnormalities	+
Shortened Limb	+
Hemidystrophy	+
Elbow malfoamation	+
Cleft palate	-
Hip Dislocation	+
Kidney Problem	-

Attach photos and Karyogram pictures

Fig 1

Fig 2a

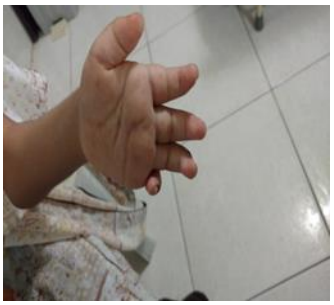

Fig 2b

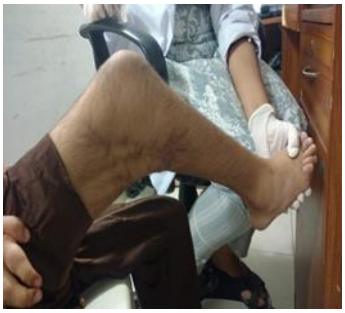

Fig 3a

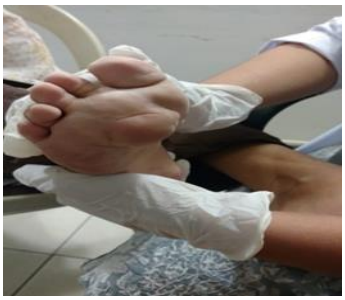

Fig 3b

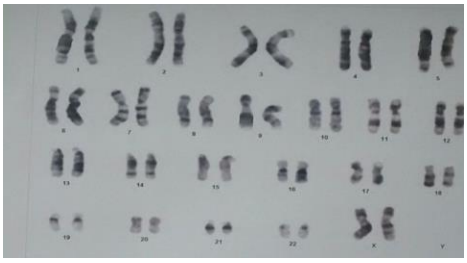

Fig 4a

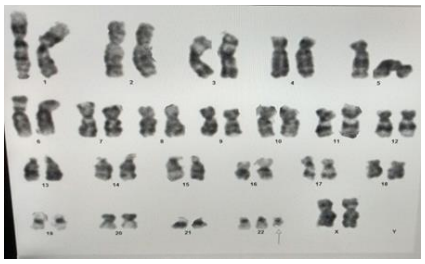

Fig 4b

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