

Pallister-Killian Syndrome- A rare genetic disorder

A Case report

Background

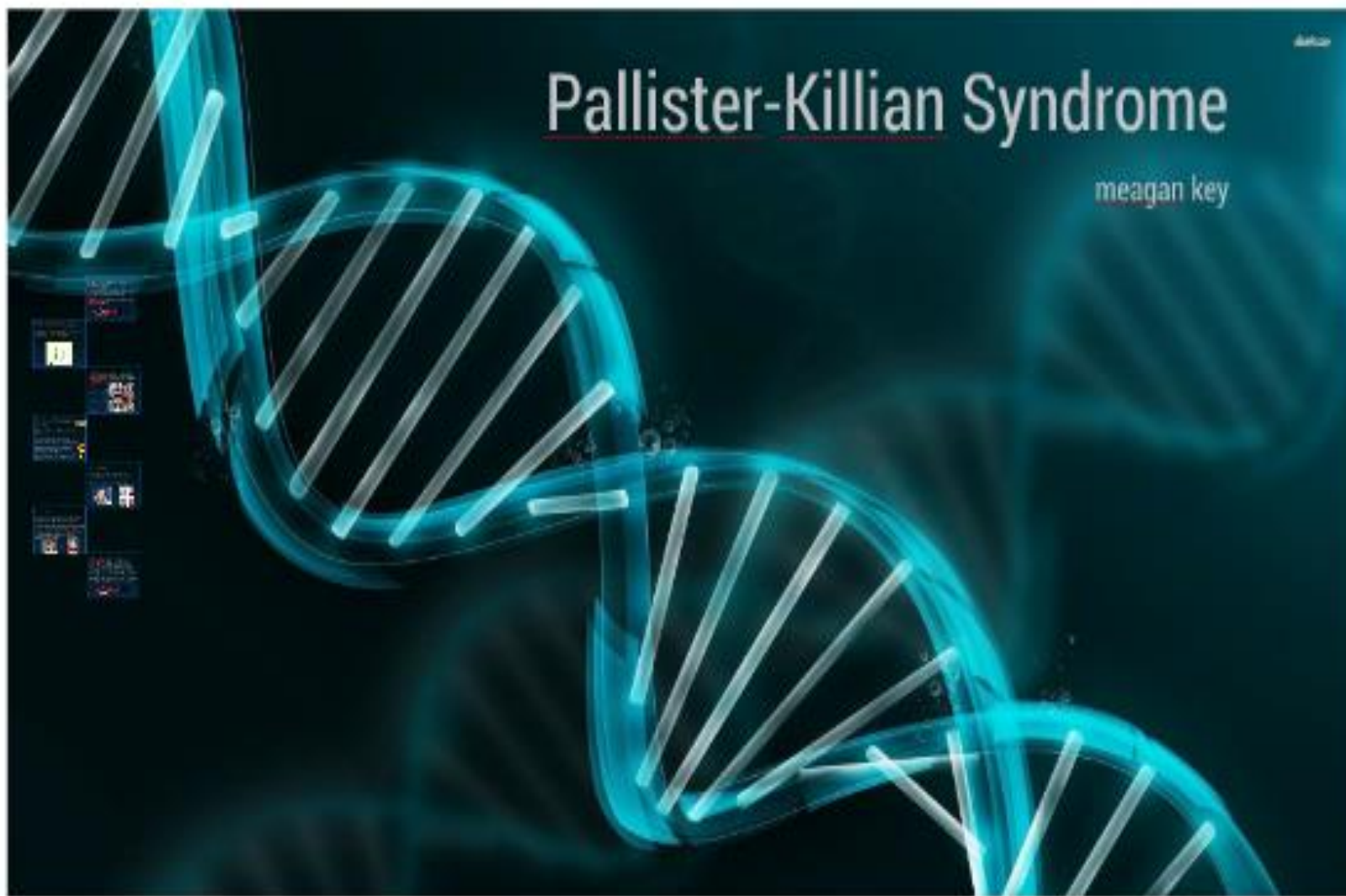
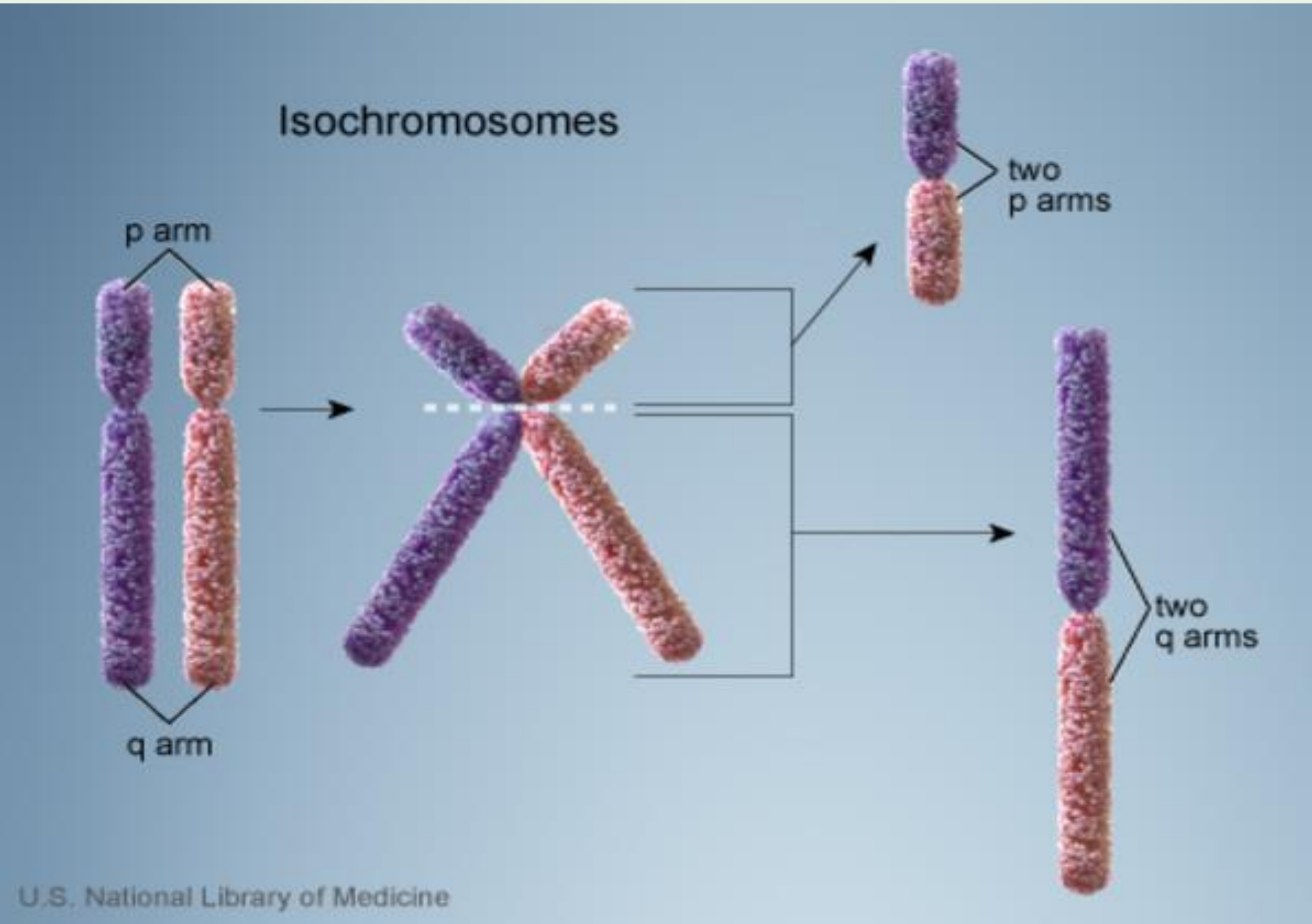
Pallister Killian Syndrome is a rare chromosomal disorder involving development in infancy and early childhood with the incidence of 1/20000. It affects multiple systems in the body including muscular hypotonia, low intelligence quotient, distinctive facial features such as high forehead, widely spaced eyes, broad nasal bridge, epicanthic folds, high arched or cleft palate and alopecia, along with cardiac anomaly, short extremities, diaphragmatic hernia, exomphalos, polydactyly and so on (5). This rare genetic disorder is caused by tetrasomy of the short arm of chromosome 12, also known as isochromosome 12p syndrome (3). This rare syndrome was first reported first By Pallister in 1977 (1) and by Teschler-Nicola and Killian in 1983 (2). The presence of 4 extra copies on short arm of chromosome 12 was identified by Pallister on skin fibroblast in 2003

Pallister-Killian Syndrome is diagnosed by karyotyping or array comparative genotypic hybridisation. Antenatal amniocentesis has low sensitivity to confirm the diagnosis (4) and this syndrome is usually confirmed via tissues with slower cell rejuvenations such as skin or buccal smear post-delivery.

Case Report

This is a case report of a 40 year old primigravida of same-sex couple conceived through intra-uterine insemination of donor sperm. An early pregnancy scan at gestation of 9 weeks and 6 days confirmed a viable pregnancy. A repeat scan at 13 weeks and 6 days gestation showed sonographic features of left sided diaphragmatic hernia with bowel and stomach involvement. The Couple were counselled for further diagnostic investigations and they proceeded with amniocentesis performed by Foetal Maternal Specialist consultant at gestation of 15weeks and 5 days. Initial PCR reported normal chromosomal and sex aneuploidies. However, culture results suggested mosaicism for 12p chromosomes – 73% normal complement and 27% structurally abnormal extra chromosomes, suggesting the clinical diagnosis of Pallister-Killian mosaic syndrome. The couple were then referred to foetal maternal specialist in Dublin tertiary centre for second opinion, as well as clinical geneticist and neonatal cardiologist.

Foetal echo suggested dextrocardia with structurally normal heart and sonographic left sided diaphragmatic hernia. Regular review by foetal biometry follow up at 21+1, 22+6, 28 week, 29+6, 30+6 were facilitated. Polyhydramnios was noted on routine ultrasound scan, with normal results of oral glucose tolerance test. Her case was discussed in multi-disciplinary team involving neonatologists, obstetricians and clinical geneticist when the plan was made for delivery in tertiary centre in Dublin for better coordination of postnatal care in Children’s Hospital Crumlin. However, premature preterm rupture of membrane was confirmed at gestation 31+5 weeks and the patient was transferred to tertiary centre for further management. A baby girl was delivered vaginally at 35 weeks gestation.



Discussion

Killian Pallister syndrome is a rare multisystem disorder with the birth incidence of 5.1 per million live births (Blyth 2015).

Identifying and diagnosing this syndrome is often very tricky antenatally through amniocentesis as reported in our case. Unfortunately, there is no specific treatment and there is a vast spectrum of disabilities requiring lifelong medical care.

In our case report, the ultimate diagnosis of Pallister-Killian Syndrome was through amniocentesis after multiple anomalies were diagnosed on ultrasound scan.

References

References:
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