BRIEF REPORT

AUTOPSY FINDINGS IN TWO CASES OF LIVEBORN TRIPLOIDY (69XXX; 69XXY)

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Summary
Two cases of triploidy (69XXX; 69XXY) in liveborn infants are described. Both cases had many of the recognized features of the triploidy syndrome. Unusual features included pulmonary immaturity, which was seen in both cases, the occurrence of cystic adenomatoid lung malformation in one case and a complete absence of Purkinje neurones in the other case.

Introduction
Triploidy is relatively common in man, occurring in about 5% of all karyotyped abortuses 1. However, it is rarely seen in liveborn infants. A recent review 2 described 54 cases, including two of mosaicism, occurring after 22 weeks of gestation. We report two cases of triploidy which survived birth. Each of these cases exhibited some hitherto undescribed features as well as many of the features described in previous reports.

Case Reports and Investigations

Case 1
A 41 year old para 6 + 4 presented at another hospital with severe pre-eclamptic toxaemia and was transferred to the National Maternity Hospital at 30 weeks gestation. Despite antihypertensive therapy, the B.P. remained at 170/120 mm Hg and proteinuria (11 g/day) persisted. A female infant was delivered by caesarean section at 31 weeks. The infant weighed 1296 g at birth and died 3 hours later.

The baby had multiple external abnormalities which included bilateral epicanthic folds, hypertelorism and microphthalmia. The bridge of the nose was flattened and the tongue bulky and protruding. The head circumference was 26 cm. General mild oedema was noted, especially in the thighs and the dorsa of the hands. Cutaneous syndactyly of the 2nd, 3rd and 4th digits of the hands and feet was present and both thenar eminences were hypoplastic. The brain weighed 1296 g at birth and died 3 hours later.

The combined lung weight was 28 g (expected weight 38 g). The cut surface of the left lung showed multiple small cysts which, on microscopic examination (Fig. 1) consisted of a proliferation of a network of terminal bronchiole-like structures variably lined by cuboidal and low columnar epithelium. The supporting stroma consisted of loose connective tissue. These changes are typical of those seen in cystic adenomatoid malformation of the lung, as described by Stocket et al. In addition, striated muscle fibres were seen running between the cysts (Fig. 2).

Karyotypes were prepared from peripheral leucocytes and revealed a 69XXX chromosome composition. Structural chromosomal abnormalities were not seen. The parents' chromosomes were not analysed.

Fig. 1 — Lung in Case I H and E stain. Note network of bronchiole-like structures of varying size, characteristic of cystic adenomatoid malformation. Magnification 10X.

Fig. 2 — Lung in Case I, PTAH stain. Note strands of striated muscle (arrow). Magnification 40X.

Case 2
The mother was a 32 year old para 1 + 1 who was admitted to the National Maternity Hospital at 30 weeks gestation in labour. A live infant of indeterminate sex was delivered and died 45 minutes after birth.

Multiple external abnormalities were present (Fig. 2). These included epicanthic folds, bilateral microphthalmia and small opacified corneas which were vertically ovoid. The bridge of the nose was flattened and the palate was cleft posteriorly. The head circumference was noticeably increased (33.5 cm). No cardiac anomaly was seen. The combined lung weight was 28 g (expected weight 38 g). The cut surface of the left lung showed multiple small cysts which, on microscopic examination (Fig. 1) consisted of a proliferation of terminal bronchiole-like structures of varying size, characteristic of cystic adenomatoid malformation. The supporting stroma consisted of loose connective tissue. These changes are typical of those seen in cystic adenomatoid malformation of the lung, as described by Stocket et al. In addition, striated muscle fibres were seen running between the cysts (Fig. 2).

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erior fontanelle. The ears were small and low-set. A small
exomphalos containing loops of small intestine was present.
Abnormalities of the external genitalia consisted of micro-
phallus and bifid scrotum. Simian creases were present on
the palms of both hands and cutaneous syndactyly of the 2nd,
3rd and 4th digits of both hands and feet was present. The
left hallux underlay, the second toe and the toenails were
hypoplastic.

The brain weighed 175 g and external hydrocephalus was
evident. Coronal sectioning of the brain revealed partial
posterior agenesis of the corpus callosum and the basal
ganglia were imperfectly formed. The cerebellum appeared
grossly normal but histologic examination revealed a complete
absence of Purkinje neurones. The other layers of the
cerebellum were normal.

The heart weighed 14 g and there was marked right ven-
tricular hypertrophy with infundibular type pulmonary stenosis.
A membranous type ventricular septal defect was present. The
lungs weighed 24 g (expected weight 36 g) and were dark
and unaerated. Histology revealed extreme immaturity with
an appearance consistent with a gestational age of 20 weeks.
The adrenals were hypoplastic (0.9 g) and the foetal cortex
was reduced. The liver, spleen and pancreas were normal
except for prominent foci of extramedullary haematopoiesis.
The thyroid and thymus were markedly reduced in weight (0.4
to 2.3 g respectively). The testes were located intra-abdom-
inally and had marked interstitial cell hyperplasia and
hypertrophy.

Microscopic examination of the eyes showed microcornea
with sclerization, congenital aniridia, ectopia lentis, malfor-
mation of the optic cup and layers, retinochoroidal coloboma,
optic disc coloboma and retinal neurone dysplasia. The
placenta weighed 800 g without cord and membranes and was
grossly normal. It was not examined histologically.

Chromosome preparations were made from peripheral
leucocytes and skin fibroblast culture and displayed a con-
figuration of 69XXY. Karyotype analysis of the parents re-
vealed normal patterns. However, a heteromorphism was
evident in the father’s C9 chromosomes (Fig. 3), one of these
chromosomes being noticeably longer than the other. A simi-
lar heteromorphism was present in two of the infant's C9
chromosomes (Fig. 4). This suggests a paternal origin for
the extra chromosome.

Discussion
The two cases reported here illustrate many of the features
of triploidy described in the literature. Thus, in Case 1,
the pregnancy was complicated by marked maternal hyper-
tension, oedema and proteinuria. This clinical condition
resembled the maternal syndrome associated with hydrops
foetalis and with maternal diabetes mellitus, hydatidiform
mole and with multiple pregnancy. It has also been noted in
association with triploidy. The enormous placenta in Case 1
showed no gross or microscopic evidence of hydatidiform
degeneration or trophoblastic proliferation. In Case II the
placenta was also enlarged, but there were no maternal
symptoms.

The finding of cystic adenomatoid lung malformation in
association with triploidy is unique, although it has been
described in association with other congenital malformation
syndromes. The presence of skeletal muscle in association