

Answers to Medical Quiz

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A1. The deformity demonstrated is phocomelia of the forearm.

A2. Thalidomide is the drug known to cause such a deformity if taken during early pregnancy.

A3. Fore-arm amputees have the opportunity to be fitted with artificial limbs. It is advised to fit children with prostheses as soon as possible to help them adjust to using such limbs. Prostheses can be just cosmetic or functional. Patients are individualised because acceptance is variable. These children need psychological support encouragement and re-enforcement to accept prosthesis as early as possible so that they adjust to the deformity and minimise psychological trauma resulting from peer scrutiny and inadvertent remarks made by these peers.

As early as 1937, FJ Browne, in his popular textbook of many editions, discussed the diagnosis of fetal abnormalities, but not their causes¹. In 1941, when the damaging effects of rubella were demonstrated, the peculiarity of the infection was emphasised but it was not taken as a general warning about vulnerability of the fetus. One long pre-thalidomide review of evidence of the effects of drugs on the fetus contained 354 references most of them about damage to the fetus and many of them published in influential and widely read journals².

Thalidomide, a sedative drug first synthesised in 1953, created one of the most dramatic disasters in the history of medicine. From 1958 the drug had been widely praised, advertised and prescribed as an unusually safe drug. It was exceptionally effective in the treatment of morning sickness in pregnant women.

Echos of the 1959 to 1961 thalidomide disaster are still with us in the form of the continuing appearance although more rarely of malformed children with phocomelia. Amelia or complete absence of a limb is an extremely rare congenital anomaly with an incidence of 0.15 per 10,000 livebirths³. There is no evidence for familial recurrence of amelia.

A case of thalidomide syndrome diagnosed by ultrasound in the 17th week of pregnancy has been reported. The pregnant woman had leprosy and received adjuvant treatment with thalidomide. The affected fetus was exposed to the drug until the 35th day of pregnancy and presented absent external ears, upper limb phocomelia and absent tibiae and fibulae. No internal organ abnormalities were noticed at autopsy⁴.

Apparently, a teratologic timetable could be fashioned retrospectively, supporting the concept that different body parts and systems have periods of vulnerability and of resistance to thalidomide, the most potent primate teratogen known. Between one out of two to one out of ten fetuses exposed at the critical development period were affected. The clinical effects are described from the literature on approximately 2500 children. Thalidomide - induced malformations are indistinguishable from sporadic non-thalidomide cases of malformations that continue to occur⁵.

Thalidomide-induced deformities, affecting 400 British children, caused various combinations of malformations. Classified by type of defect and whether confined to upper and / or lower limbs, the malformations can be treated on a rational basis. The treatment from infancy onward includes the fitting of carbon dioxide gas - powdered, upper limb prostheses, initially in their simplest form and finally in the most sophisticated form. For a baby with bilateral lower phocomelia, a special 'sitting socket' was devised to enable sitting in an upright position⁶.

REFERENCES

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