May be associated with one or more cutaneous manifestations:

- Skin depression or dimple
- Port-wine angiomatous nevi
- Dark tufts of hair
- Soft, subcutaneous lipomas

May have neuromuscular disturbances:

- Progressive disturbance of gait with foot weakness
- Bowel and bladder sphincter disturbances

Prenatal Detection

It is possible to determine the presence of some major open NTDs prenatally. Ultrasonographic scanning of the uterus and elevated maternal concentrations of alpha-fetoprotein (AFP, or MS-AFP), a fetal-specific gamma-1-globulin, in amniotic fluid may indicate anencephaly or myelomeningocele. The optimum time for performing these diagnostic tests is between 16 and 18 weeks of gestation before AFP concentrations normally diminish and in sufficient time to permit a therapeutic abortion. It is recommended that such diagnostic procedures and genetic counseling be considered for all mothers who have borne an affected child, and testing is offered to all pregnant women (American College of Obstetrics and Gynecology Committee on Practice Bulletins, 2007). Chorionic villus sampling is also a method for prenatal diagnosis of NTDs; however, it carries certain risks (skeletal limb depletion) and is not recommended before 10 weeks of gestation (Simpson, Richards, and Otano, 2012).

Therapeutic Management

Management of the child who has a myelomeningocele requires a multidisciplinary team approach involving the specialties of