Schinzel syndrome, also known as ulnar-mammary syndrome, is a rare inherited disorder characterized by abnormalities of the bones of the hands and forearms in association with underdevelopment (hypoplasia) and dysfunction of certain sweat (apocrine) glands and/or the breasts (mammary glands). Abnormalities affecting the hands and/or forearms range from underdevelopment of the bone in the tip of the fifth finger (hypoplastic terminal phalanx) to underdevelopment or complete absence of the bone on the outer aspect of the forearm (ulna). In some cases, a diagnosis of Schinzel syndrome may be made at birth based upon a thorough clinical evaluation, the identification of characteristic physical findings, and specialized imaging techniques. Such imaging studies may be conducted to confirm and/or characterize bone abnormalities affecting the fingers, hands, wrists, and/or arms; certain genital abnormalities (e.g., bicornate uterus in females, cryptorchidism in males); and/or other malformations (e.g., pyloric stenosis, inguinal hernia). Specialized tests may also be conducted to detect and verify dysfunction of certain sweat (apocrine) glands and/or the mammary glands in affected females. In addition, in some cases, additional testing may be conducted (e.g., echocardiograms, electrocardiograms, cardiac catheterization, specialized x-ray studies, etc.) to detect the presence of and/or characterize ventricular septal defects, which have been reported in one family with the disorder.