The Rabenhorst-Syndrome
A Cardio-Acro-Fascial Syndrome*

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Abstract. A syndrome, apparently not yet described, with following manifestations is presented: 1. Ventricular septal defect and pulmonary stenosis. 2. Narrow face with micrognathia, high and narrow nose with prominent septum, microstomia, attached earlobes. 3. Minor malformations of hands and feet. The inheritance appears to be autosomal dominant.

Key words: Autosomal dominant malformation syndrome — Ventricular septal defect — Pulmonary stenosis — Facial malformations — Anomalies of hands and feet.


Introduction

The incidence of congenital heart malformation among live-born children is between 0.5 and 1% (Higgins, 1965; Campbell, 1965; Richards et al., 1955; Jørgensen et al., 1971). The reported occurrence of associated extracardial malformations varies from 9 to 43% (Campbell 1965; Quittek, 1968; Heck, 1955); information on these malformations reflecting size and age group of the sample, the applied definition of the term malformation, and whether the material was gained clinically and/or by autopsy. Emeritt claims that recognizable syndromes — including the chromosomal syndromes — account for only 5% of these cases. Among the recognizable syndromes there are only a few with a primary heart defect (in contrast to those with a heart affection being secondary to the basic defect of the disease, e.g. Hurler's and Marfan's syndrome) and apparently simple Mendelian inheritance: including Holt-Oram and

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Leopard syndrome, dominantly inherited, and the Ellis van Creveld and Meckel syndrome, recessively inherited.

The aim of this paper is to present a syndrome, apparently not yet described, that most probably is passed on in an autosomal dominant mode of inheritance. The "leading" manifestation appears to be a heart defect, which is accompanied by multiple other anomalies, particularly of the face and extremities.

Case Reports

The proposita, pedigree No. IV, 2 (Fig. 1) was born on Sept. 19, 1969 after an uneventful pregnancy. Delivery had to be initiated because of postmaturity but was otherwise normal. Birth weight and length were within normal limits. Because of prolonged cyanosis she got oxygen for 1 day. Later a heart murmur was noted and swelling of the feet appeared; she was referred to us for these reasons at the age of 6 weeks.

Fig. 1. Pedigree of the R family