遺伝性心疾患　　Mar 20　2012

ECG localization of bypass tracts ;

The “key” is to look for the leads with the negative delta wave,
because the negative delta wave “points” to the bypass tracts.

Mild valvar PS（ RVp＜ 50% systemic,　gradient 35-40mmHg ）　（Nugent EW　1977）
These patients have a normal hemodynamic response to exercise and
have good long-term outcomes.

Of the 261 patients followed medically, all age groups included a few patients
in whom a low initial gradient （ 40mmHg or less ）decreased through the years.

30% of patients first diagnosed at less than 1mo progress to moderate to severe obstruction. These patients should have w/u at 6mo since progression does occur in the first 6months, thereafter very uncommon.
No need for Endocarditis Prophylaxis

ASD　　paternal disomy 14 ; maternal disomy 16
microdeletion- 1p36 ; 5q35 ; 6p25 ; 9q34 ; 15q24-q26

spontaneous closure of ASDs
diagnosed in the first 3 mos of age　（HSC Toronto　1993）

ASD ＜ 3mm	32/32	100%
3 to 5mm	39/45	87%
5 to 8mm	16/20	80%
≧ 8mm	0/4	0%
total	87/101	87%

ASD＜ 3mm : No need for 2nd w/u since 100% of these defects will close by 18 mos

ASD 3-5mm : 2nd w/u at 12mos

ASD 5-8mm : 2nd w/u at 15mos

ASD ≧ 8mm may not have spontaneous closure and
may need intervention

ECD	maternal disomy 16 microdeletion- 3p25 ; 8p23 ; 10q22
CoA/IAA	mosaic chromosome 16 ; maternal disomy 16 microdeletion- 6p25 ; 10p15 ; 22q11 microduplication- 22q11.2
dextrocardia	microdeletion- 6p27
right aortic arch	microdeletion- 12p13 ; 22q11
DORV	microdeletion- 8p23 ; 10p15
TOF	microdeletion- 5q11.2 ; 9q34 ; 10p15 ; 22q11 microduplication- 22q11.2
TGA	microdeletion-10p15 ; 22q11
HLHS	microdeletion- 1p36 ; 8p23 ; 11q23-qter ; 14q23 ; 15q24-q26 20q11 microduplication- 22q11.2
PDA	maternal disomy 2; paternal disomy 6 ; paternal disomy 14 microdeletion- 5q35 ; 6p25 ; 9q34
PS	microdeletion- 5q35 ; 9p13 ; 20p
TAPVC	microduplication- 22q11.2
VSD	mosaic trisomy 10 ; paternal disomy 10 ; maternal disomy 16 microdeletion- 1p36 ; 1q44-qter ; 4q35 ; 5q35 ; 6p25 9q34 ; 10p15 ; 17q25 ; 19p13 ; 22q11
HCM	caused by gene mutations 1q23 ; 2q31 ; 3p21 ; 11p11 ; 14q12 ; 15q14

Inherited muscle diseases with cardiac involvement
autosomal dominant

myotonic dystrophy

DM1	19q13.2-q13.3
DM2	3q13.3-q24

limb girdle muscular dystrophy

LGMD1A	5q31
LGMD1B	1q21.2
LGMD1C	3p25
EDMD	1q11-q21
FSH dystrophy	4q35
Bethlem myopathy	2q37 ; 21q22

desminopathy

primary	1p35-p36
MFM	2q35