Introduction:
In 1968 a cytogenetics laboratory was established at the University Hospital, Reykjavik and has since then served as the only chromosomal laboratory for all hospitals and physicians in Iceland. Our current aim was to study the physical features, congenital anomalies and various clinical outcomes in Icelandic females, diagnosed with Turner syndrome (TS) for the period of 1968 – 2012.

Methods:
Data was obtained from hospital records, from all pediatric endocrinologists in Iceland and the cytogenetics laboratory making this a nationwide retrospective population study.

Results:
A total of 51 females were diagnosed with TS during the 45 year period with an average yearly incidence of 1 per 2585 liveborn females. Average birthweight was 3028 grams. Clinical features were webbed neck (47%), shield chest (45%), low hairline (45%), cubitus valgus (27%), hyperconvex nails (25%) and short fingers (25%). Pedal hydrops was seen in 25% of cases at birth. Sixteen percent had bicuspid aortic valve, 12% coarctation of the aorta and 14% had horseshoe kidney. The most common late complication was recurrent acute otitis media (55%), followed by high blood pressure (29%) and various thyroid problems in 24% of cases. Three girls (6%) were diagnosed with type 2 diabetes and 1 girl had idiopathic thrombocytopenic purpura (ITP). Twenty four girls received growth hormone (GH) treatment. Eighteen have completed treatment. Average final height of girls who started GH before 1992 was 147,9 cm (-3,1SD), but for girls started on GH after 1992, 156,5 cm (-1,8 SD). Eight girls had signs of spontaneous puberty but only 1 completed pubertal development resulting in unaided pregnancy.

Conclusion:
The clinical features and the most common clinical complications are similar to what has been described in the other nordic countries. Improvement in growth hormone treatment and sex hormone replacement has resulted in improved height during the last two decades.